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Inside the Institute

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Racial Inclusivity in Neuroscience: Rodriguez-Romaguera Shares Lessons Learned in Mentor-Mentee Relationships

Jose Rodriguez-Romaguera, PhD

Diversity is the driving factor for scientific discovery. However, racial diversification efforts among researchers have fallen short. Publishing in Neuron, Jose Rodriguez-Romaguera, PhD, assistant professor in the UNC Neuroscience Center and Assistant Director of the Intellectual and Developmental Disabilities Research Center (IDDRC) at the Carolina Institute for Developmental Disabilities, authored a perspective on how critical mentor-mentee relationships are for the next generation of neuroscientists, especially for those who come from racially underrepresented groups like himself.

In the piece, Rodriguez-Romaguera and his former mentor, Gregory Quirk, PhD, a supervising scientist at the University of the Philippines-Manila, formerly a professor at the University of Puerto Rico School of Medicine, reflect on lessons learned from their cross-racial mentor-mentee relationship that could apply to many researchers today. The perspective addresses boundaries of mentor-mentee relationships, lab environments that value caring and inclusivity, and strategies to overcome impostor syndrome.

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"I did not perceive myself as a scientist prior to joining the Quirk Lab because most scientists did not look like me," said Rodriguez-Romaguera. "In fact, my own mentor did not look like me! However, my mentor, Greg, who "looked" like a scientist telling me I could also "be a scientist" was a powerful motivator and started my journey to become a neuroscientist."

To create racially inclusive environments, this perspective acknowledges areas of focus and questions to address in order to be more effective in mentoring trainees from underrepresented and historically excluded backgrounds. Fostering a mentee's sense of belonging can ignite a positive attitude in academic pursuits leading to career success. Yet, the perspective explains how the imposter syndrome – one's self-doubt in intellect or skills – can make mentees feel incapable of belonging in the research realm. It is in this instance where Rodriguez-Romaguera urges mentors to allocate time in being accessible to help trainees form their scientific identity.

"As a graduate student, I used to suffer from this type of impostor phenomenon until my mentor asked me, 'Do you prefer to fail, or would you rather succeed and leave your impostor identity behind?' While I was horrified that my mentor could think that I was failing on purpose, I realized that there were unconscious processes preventing me from succeeding in an environment where I thought I already belonged," said Rodriguez-Romaguera.

Acknowledging the mentee as the whole person with proper caring, understanding and boundaries is another recommendation for establishing a collaborative mentor-mentee relationship. Rodriguez-Romaguera states that personal issues and scientific issues can be successfully compartmentalized between a mentee and mentor. Personal problems can affect a mentee's work professionally, and it's important for the mentor to recognize the challenges these can bring to both academic and nonacademic life. On the other hand, the piece stresses the importance of balancing boundaries between the mentor and mentee. Mentoring can lead to a friendship which poses the risk of hurting the purpose of the relationship, and it can spiral the mentee into establishing unnecessary dependence.

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Racial Inclusivity in Neuroscience: Rodriguez-Romaguera Shares Lessons Learned in Mentor-Mentee Relationships *continued*

"My mentor, Greg, jolted me late one afternoon by saying, 'I may be friendly, but I'm not your friend, I'm your mentor,'" said Rodriguez-Romaguera. "While inconvenient to hear, this can allow the mentee to relax, knowing that the mentor will not lose sight of the mentee's professional needs. Establishing boundaries by both the mentor and the mentee are very important to mentoring scientists so the relationship maintains its focus on the science and the career progression of the trainee."

When it comes to focusing on productivity or focusing on a caring, personal tone, Rodriguez-Romaguera says both are necessary for the mentee. Additionally, he says trainees from underrepresented backgrounds may need more caring initially than non-underrepresented trainees because of the additional work needed to overcome racial stereotypes. From the positive lab environment to participating in close-knit group activities, building cohesiveness between a mentor and mentee can strengthen career aspirations, especially as a mentee moves into postdoctoral training and eventually become mentors in their own labs.

NC State Developmental Centers Community Interest Project

The CIDD team traveled to Murdoch Developmental Center earlier this year. Back row L to R: Eyram Bossiade (CIDD, LEND MSW trainee), Joy Solomon (Murdoch, Director of Programs), Morgan Parlier (CIDD, Director of Family Support Services). Second row L to R: Isabella Russo (CIDD, LEND MSW trainee), Lauren Winfrey (CIDD, LEND MSW trainee), Anna Ward (CIDD, Director of Advocacy and Inclusion), Annamae Giles (UNC Cares Project Manager). Front row L to R: Marcia Roth (Systems Liaison with CIDD), Dava Hunt (Murdoch, Director of Social Work), Kim M (Murdoch).

The Carolina Institute for Developmental Disabilities (CIDD) in collaboration with UNC Cares, an established collective impact initiative of the University of North Carolina School of Social Work, are partnering to explore the understanding, needs, and interest in community living by residents within the State Developmental Centers (SDC) and their legally representative parties (LRP)/legal guardians.

The population of focus for this project are longtime residents of the SDCs (Murdoch Center, J. Iverson Riddle Center, Caswell Developmental Center), since 2011 or before. Beginning in 2012, anyone entering an SDC was required to have a memorandum of agreement (MOA), which included a transition to community plan developed in conjunction with their Local Management Entity/ Managed Care Organization (LME-MCO). A consistent goal in NC has been to assure individuals are provided education and choice regarding least restrictive settings for community living.

The U.S. Supreme Court decision Olmstead v. L.C., 119 S.CT. 2176 (1999) mandated that states serve individuals with developmental disabilities in the least restrictive environment as specified in the Americans with Disabilities Act (ADA) of 1990. In December 20, 2021, North Carolina published The North Carolina Olmstead Plan.

The Division of State Operated Healthcare Facilities (DSOHF) has contracted with the UNC Team to design and implement the "SDC Community Interest" project. The UNC team will build capacity through the establishment of an SDC Project Advisory Council. The Council will consist of 12-15 members representing Universal Design content area experts, disability advocacy organizations, MCOs, self and family advocates, and LRPs to advise the project team on:

- Education materials for eligible SDC residents and their LRPs to promote person-centered planning and Supported Decision-Making (SDM)
- Application of Universal Design principles to modify existing survey tools to maximize individual participation, with use of assistive and augmentative communication (AAC) and proxies, as indicated
- Interviewer training protocols for surveying individuals with I/DD and/or complex communication needs
- Interview process to reach ~650 residents of the 3 SDCs and their LRPs
- Analysis of the data and the report to DSOHF and diverse audiences
- Contributions to the "SDC Community Interest" project and outcomes evaluation

UNC's Whole Brain Health Program
Transition for Youth with Autism and/or Epilepsy

The University of North Carolina has been awarded $2.2 million from the Health Resources and Services Administration's Maternal and Child Health Bureau (HRSA MCHB) to be one of eleven National Transition for Youth with Autism (4 grants) and/or Epilepsy (7 grants) Demonstration Projects. CIDD community members who contributed to the proposal and will be involved in the project include Diana Cejas (PI) and Anne Harris (Co-PI) alongside Marcia Cordova Roth, Julie Williams-Swiggett, Morgan Parlier, Anna Ward, Kenneth Kelty, and Margaret DeRamus. Other involved university partners include neurologist Dr. Linn Liu and psychologist Dr. Blaise Morrison.

This five-year project aims to improve national, state, and local/community-level frameworks that support a successful transition from child to adult serving systems for youth with autism and/or epilepsy who have complex health and social needs and require a higher level of family support and coordination. The goal of UNC's Whole Brain Health Program (WBHP) is to improve transition outcomes, quality of life and well-being for youth ages 13-26 with Epilepsy and co-occurring intellectual and/or developmental disabilities with or without mental/behavioral health diagnoses, and their families/caregivers as they transition from child to adult systems including pediatric to adult health care, education, employment, and community living. To better serve the many individuals with epilepsy and intellectual developmental disabilities (I/DD) and complex care needs requiring family support and coordination, the UNC Department of Neurology will partner with the Carolina Institute for Developmental Disabilities (CIDD), the NC Division of Child and Family Wellbeing/CYSHCN Program, and diverse state and community stakeholders including disability and family organizations and individual patients/self-advocates. Funding will expand capacity for a comprehensive, transdisciplinary WBHP Transition Clinic, hire family navigation and peer support staff, train and mentor pediatric and adult healthcare providers, and implement systems science approaches to improve coordination and alignment of existing and emerging transition resources for NC youth with co-occurring epilepsy and intellectual and/or developmental disabilities.

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Kenneth Kelty
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Congratulations to Mark Shen, Recipient of the 2024 Hettleman Prize

Mark Shen, PhD

Congratulations to this year's Hettleman Prize winners! Dr. Mark Shen was chosen as one of these five early-career faculty exemplifying groundbreaking and innovative research along with future career promise. Shen is a developmental neuroscientist, Assistant Professor of Psychiatry, and Investigator in the UNC Neuroscience Center and Carolina Institute for Developmental Disabilities. He discovered an early biomarker for autism — the presence of excessive cerebrospinal fluid (CSF) volume in the brain — that is detectable by 6 months of age in certain babies, which is two years before they develop autism. His research since then has focused on how CSF cleans the brain of neuroinflammation and how problems with that process can lead to the development of autism in the first years of life. He has led the world's literature in this field.

Shen is now PI of a five-year, NIH-funded study of early CSF pathophysiology, the brain's lymphatics, and its downstream effects on brain development, using neuroimaging in infants with neurodevelopmental disabilities (NDDs) and mechanistic studies in corresponding mouse models of these NDDs. His NIH grant is the first study of brain lymphatics in children. Shen has published 40 peer-reviewed papers, including eight that were recognized by the NIH as finalists for "the most significant advances in autism research." He was also named the Early-Career Investigator of the Year by the International Society for Autism Research.

Behavioral symptoms of autism emerge in the latter part of the first or second years of life, and therefore diagnosis is not currently made until 2-4 years of age. Shen has demonstrated that enlargement of CSF volume emerged in the first six months of life in infants who later developed autism, allowing for the future possibility of a new, earlier method of diagnosis. In 2018, Shen founded the clinical trials program within the Carolina Institute for Developmental Disabilities, which tests novel genetic therapies in children with NDDs. The over-arching goal of Shen's research is to identify early biological markers and therapeutic targets for NDDs including autism, fragile X syndrome, Down syndrome, and Angelman syndrome. His research is unique because it is fully translational between preclinical models, clinical research, and clinical trials.

"I am beyond humbled and honored by this prestigious award," Shen says. "I view this as a team award, as I've been very fortunate to work alongside clinicians and basic scientists, from physicians to clinical trialists, in order to make discoveries that will hopefully increase the quality of life for children with autism and developmental disabilities."

Gabriel Dichter Awarded NICHD Grant to Study Brain-Immune System Connections in Autism

Gabriel Dichter, PhD

Kaitlin Cummings

Gabriel Dichter, Associate Director for Research at CIDD, has been awarded an exploratory grant from NICHD to study linkages between brain functioning and the immune system in autism. The project builds on pilot data collected by Kaitlin Cummings, a clinical psychology graduate student in Dichter's lab. In addition to Cummings, collaborators on the project include Keely Muscatell (Psychology and Neuroscience), Alana Campbell (Psychiatry), Kinh Truong (Biostatistics), Katherine Meltzoff (UC-Riverside), and Bennett Wood, MSW, a neurodivergent practitioner and former UNC LEND trainee. The project will use the EEG resources of the UNC Intellectual and Developmental Disabilities Research Center.

It has long been recognized that individuals with autism are characterized by differences in how the brain processes social rewards. Additionally, several avenues of research indicate that autism is characterized by heightened inflammatory responses. Research by Muscatell has shown that inflammation, the body's response to physical and psychosocial stressors, impacts social behavior as well as brain circuits that process rewarding information. The goal of this new project is to see if brain function, measured with EEG during social reward processing, is associated with measures of inflammation derived from an analysis of blood levels of inflammatory cytokines in autistic individuals, and how these impact social functioning and quality of life. Additionally, because a portion of study participants have participated in a companion PET study at UNC, supported by a Neurospark Award from the UNC Neuroscience Center, the project will also investigate the correspondence between EEG, blood-based markers of inflammation, and PET measures of neuroinflammation.

Let Sleeping Babies Lie: Scientists Highlight Negative Impacts of Sleep Disruption on Early Brain Development

Sleep deprivation in adults has long been proven to cause long-term mental and physical health issues, including, but not limited to, weakened immune systems, weight gain, depression, and increased risk of dementia. But why does lack of sleep have such steep consequences? Sleep actually plays a crucial role from the moment we are born. As babies, our brains are still forming the ends of neurons, called synapses, that are important in learning, attention, working memory, and long-term memory. Sleep allows these neurons to develop and connect with one another, establishing brain functions for the remainder of life. If this delicate, but important process was to be disrupted either through constant waking or separation anxiety, it could have lasting effects on the brain and behavior.

Now, a new study led by Sean Gay, a graduate student in the lab of Graham Diering, PhD, assistant professor in the Department of Cell Biology and Physiology at the UNC School of Medicine and member of the CIDD, has given us more insights into how sleep loss during early life impacts key parts of brain development – and how it can also increase one's risk for developing autism spectrum disorder (ASD). Their findings were published in the Proceedings of the National Academy of Sciences. "The unique effects of sleep loss during development are largely unexplored," said Diering. "Our data show that babies and children are more vulnerable to the negative effects of sleep disruption. We also found that sleep loss during this crucial period of time can negatively interact with underlying genetic risk for autism spectrum disorder."

Graham Diering, PhD

Sleep Disruption and Autism

Sleep issues are an important early indicator of brain growth issues and other neurodevelopmental disorders, such as ASD, attention-deficit hyperactivity disorder, and intellectual disability. Sleep disruption has been noted in >80% of people with ASD, but whether sleep disruption is a cause or consequence of ASD is largely unknown. Diering has long studied how sleep strengthens synapses over time – a process termed synaptic plasticity – and how lack of sleep can contribute to cognitive and neurodegenerative disorders. If researchers could better understand the links between sleep and ASD, researchers and physicians also could make earlier diagnoses and come up with new treatment strategies for the disorder. In 2022, the Diering lab sought to understand if sleep disruption during early life could interact with underlying genetic risk for ASD to cause long-lasting changes in adult behavior. Using mouse models, researchers found that sleep disruption during the third week of life (similar to age 1-2 in humans) caused long-lasting deficits in social behavior in male mice that were genetically vulnerable for having ASD.

A Study on Sleep Rebound

The Diering lab wanted to explore these findings further, this time diving into how adult and developing mouse models compensate for sleep loss. Using specialized mouse houses with highly sensitive sensors, researchers were able to carefully track mouse movements and breathing, allowing the researchers to keep score of wake and sleep states. Researchers showed that when the adult mouse models lost a significant amount of sleep, they compensated for it by increasing sleep later during their regular active hours. Termed "sleep rebound", this response allowed the adults to "make up" for lost sleep.

The younger mice; on the other hand, lacked sleep rebound entirely. This confirmed the researcher's hypothesis that the younger mice might be more susceptible to the harmful effects of sleep deprivation. Researchers also noted that sleep deprivation in young mice completely impaired their performance in a learning memory task, whereas adults were far more resilient after sleep loss.

Next, the lab shifted its attention to the effects of sleep and sleep deprivation on neuronal synapses, which mediate communication between neurons and are the main location for memory formation and storage. They are also well-studied for their pivotal role in benefiting sleep health. Researchers performed a number of molecular analyses to look at how sleep deprivation affects synapses. Using cutting-edge protein analysis, they were able to map the protein composition and biochemical changes that affect synapses. The analysis showed that sleep deprivation in young mice, but not adults, strongly affected synapse formation, a key aspect of brain development. "This now provides one of the largest and most comprehensive datasets to examine the molecular effects of sleep loss across the lifespan," said Diering.

Future Treatment Avenues for Autism

An ongoing mission of the lab, informed through the molecular work of this current study, is to develop next generation sleep-based medicines that could be used in children. Instead of acting as a sedative, they hope to create a drug that can target synapses to restore sleep function, rather than altering sleep behavior itself. "Development is not something that one can go back and do again," said Diering. "Sleep is important for the entire life and especially during development. Understanding what we know now will place greater emphasis on understanding sleep issues in ASD and could lead to an important therapeutic avenue to treat ASD and other developmental conditions."

The CIDD T32 Postdoctoral Research Training Program

Drs. Mark Shen and Ben Philpot will work with a talented group of six post-doctoral research fellows in our training program on neurodevelopmental disorders research: **Scott Albert, Samuel (Sam) Barth, Marissa DiPiero, Crisma Emmanuel, Jieun (Esther) Park, and Joshua Rutsohn**. We are thrilled to have these talented postdoctoral fellows in our interdisciplinary program, and would like to introduce the fellows and the T32 Co-Directors to the CIDD community.

Dr. Mark Shen is a developmental neuroscientist, Assistant Professor of Psychiatry, and investigator in the Neuroscience Center and CIDD. He is also the founding Co-Director of the CIDD Clinical Trials Program. Dr. Shen earned his PhD from the University of California-Davis MIND Institute with David Amaral and completed the CIDD T32 postdoctoral fellowship with Joe Piven. Dr. Shen has pioneered research on the early brain development and cerebrospinal fluid physiology in autism and other neurodevelopmental disorders including fragile X, Down syndrome, and Angelman syndrome. His research identifying early brain markers of developmental disabilities has resulted in >40 publications and multiple awards including the Early-Career Investigator of the Year by the International Society for Autism Research and the Hettleman Prize. As a former T32 fellow himself and then a T32 faculty mentor since 2017, Dr. Shen is the ideal fit to co-lead our T32 training program with Dr. Philpot.

Dr. Ben Philpot has been the Director of the CIDD T32 postdoctoral training program since 2023, having previously served from 2009-2023 as the Associate Director. Dr. Philpot is a Kenan Distinguished Professor in the Department of Cell Biology & Physiology. He earned his Ph.D. at the University of Virginia and performed a postdoctoral fellowship in the laboratory of Dr. Mark Bear at M.I.T. and Brown University. He is the Associate Director of the UNC Neuroscience Center and a member of the Carolina Institute for Developmental Disabilities. Dr. Philpot seeks to understand the pathophysiology underlying monogenic neurodevelopmental disorders, and he is developing small molecule and gene therapies to treat these disorders. His research focuses on early-stage development of treatments for Pitt-Hopkins, Dup15q, and Angelman syndromes. Dr. Philpot has >100 peer-reviewed scientific publications and has won multiple awards for his work in neurodevelopmental disorders.

Dr. Scott Albert received his PhD in Biomedical Engineering from Johns Hopkins University working with Dr. Reza Shadmehr. His research examines resilience in motor circuits across humans and mice. As a UNC CIDD T32 Fellow, Dr. Albert will work with Dr. Adam Hantman, in examining whole-brain maps of motor activity in mouse models of Angelman syndrome and Dup15q syndrome. He will use fMRI and electrophysiology to identify how UBE3A expression regulates the adaptability and robustness of motor control circuits.

Dr. Samuel Barth received his PhD in Neuroscience from Wake Forest University working with Dr. Kimberly Raab-Graham. His graduate research examined how mTOR dysregulation affects GABAergic synaptic formation and elimination in a neurodevelopmental disorder called Tuberous Sclerosis Complex (TSC). As a UNC CIDD T32 Fellow, Dr. Barth will examine the molecular contributions to Dup15q and Angelman syndromes while working with Dr. Ben Philpot. His research will disambiguate how differential expression of Ube3A contributes to Dup15q and Angelman syndrome pathology.

Dr. Marissa DiPiero received her PhD in Neuroscience from the University of Wisconsin-Madison under the mentorship of Dr. Doug Dean. Her research focuses on the development and utilization of diffusion MRI methodologies to study brain microstructure across the lifespan and uncover differences in brain-behavior relationships associated with autism. As a UNC CIDD T32 fellow, Dr. DiPiero will work with Dr. John Gilmore to investigate individual differences in the developmental trajectories of brain-behavior relationships in children at an increased risk for neurodevelopmental conditions, including autism and schizophrenia.

The CIDD T32 Postdoctoral Research Training Program *continued*

Dr. Crisma Emmanuel received her PhD in Nursing from UNC at Chapel Hill working with Dr. Hudson Santos. Her research examines socio-environmental factors that affect child development and health among children at high risk of developing a neurodevelopmental disorder. She is currently a T32 postdoctoral fellow working with Dr. Michael O'Shea on examining environmental factors that affect the health and development of extremely preterm born children. As a T32 fellow at the CIDD, Dr. Emmanuel is evaluating biosocial factors affecting the well-being of children with a neurodevelopmental disorder.

Dr. Jieun (Esther) Park received her PhD in Cell Biology from Duke University with Dr. Michel Bagnat. Her PhD work focused on the development of specialized intestinal cells important for protein absorption. For her postdoc, she decided to switch gears and focus on neuroscience with the eventual goal to better understand the connection of brain and gut development. As a UNC CIDD T32 fellow, Dr. Park works with Dr. Jason Stein to investigate the underlying molecular mechanisms leading to cortical surface area expansion in autistic individuals early in life using brain organoids as a model system.

Dr. Joshua Rutsohn received his DrPH in biostatistics from the Gillings School of Public Health at UNC-Chapel Hill under the guidance of Dr. Young Truong. His research encompasses the estimation of latent variables from longitudinal and time-series data. As a UNC CIDD T32 Fellow, Dr. Rutsohn works with Dr. Truong to construct latent classes from signal processing and neuroimaging data. His research aims to develop new methods to model heterogeneity inherent to neurodevelopmental data.

Work Together NC—Working Together from Possibility to Opportunity

Work Together NC (WTNC) is a community collaboration that brings together self-advocates, families, employers, state agencies, and service providers to improve access to opportunities and resources focused on the transition to adulthood for people with intellectual and developmental disabilities.

WTNC has created accessible action planning tools to aid in planning for the transition to adulthood. The goal is for these tools to be utilized collaboratively by persons with an intellectual or developmental disability (IDD), family members, IEP team members, and/or an adult service providers. The first category is about what opportunities are available after high school. This includes action plans on employment, postsecondary education, and community living. The second category includes topics that address transitioning smoothly to adulthood. This includes action plans on healthcare, adult services, financial well-being, transportation, and IEP. The final category includes action plans on topics like self advocacy and supported decision making/guardianship. This project is supported, in part by grant number 90DNCE0006 from the U.S. Administration for Community Living, Department of Health and Human Services, Washington, D.C. 20201.

New Inclusive Postsecondary Education Programs for Students with IDD in the UNC System

THE UNIVERSITY OF NORTH CAROLINA SYSTEM

State Legislature appropriated funds to support 3 new programs in the UNC System, and UNC Chapel Hill received foundation funds and administrative approval to begin offering HEELS UP during the academic year in addition to the summer program.

UNC Chapel Hill - HEELS UP

- First cohort will be in Spring 2025 (applications are already closed)

UNC Wilmington - Inclusive Future Programs

- First cohort will be in Fall 2025 (applications due 3/1/25)

NC State - Elevate

- First cohort will be in Spring 2025 (applications are already closed)

North Carolina Central University-TBD

- First cohort expected in Fall 2025

Welcome to Access to Achievement programs!

The NC legislature also appropriated funds to the NC Community College System to provide grants to 15 out of NC's 58 technical and community colleges. Many of the programs are in development but will start in 2025. The following campuses received awards:

NC COMMUNITY COLLEGES
CREATING SUCCESS

Richmond	Wilkes	James Sprunt
Robeson	Alamance	Johnston
Sandhills	Asheville-Buncombe	McDowell
South Piedmont	College of the Albemarle	Brunswick
Stanly	Guilford	Catawba Valley

2024 State of The Art (SOTA) Conference

The State of the Art in Postsecondary Education for Students with ID (SOTA) Conference was recently held in Chapel Hill, North Carolina. On October 28th, 37 colleges from around the country were represented at the inclusive college fair. The NC community college scene in particular has significantly grown, and their presence was felt. On October 29th and 30th, we had over 400 conference attendees from across the United States and around the world.

Conference speakers included a keynote from Rehabilitation Services Administration (RSA) Commissioner, Dante' Allen, national faculty and staff from postsecondary education initiatives, parents, self-advocates, and other experts-sharing effective practices and leading group discussions. Our own Kenneth Kelty moderated a panel of students from the NC inclusive postsecondary programs at Western, UNC-Greensboro, and Appalachian State.

CIDD Disability Advocate, Kenneth Kelty (seated far right) moderates SOTA plenary panel of students from the NC inclusive postsecondary programs at Western, UNC-Greensboro, and Appalachian State.

There was also a parallel Student Leadership Conference (SLC) that brought current and prospective college students into the conversation with opportunities to learn useful tools for the transition to college life, expand their skills as student advocates and leaders, and meet peers from across the country. Together, through both conferences, we raised $2500 for schools affected by Hurricane Helene in Western Carolina.

Pictured right: Representatives from the NC Community College System attend the 2024 SOTA Conference. SOTA provides opportunities for colleges, universities, researchers, program staff, parents and self-advocates to learn about the current state of research and practice in the field of inclusive postsecondary/higher education, and to network with each other.

Researchers Identify Potential Treatment for Angelman Syndrome

Angelman syndrome is a rare genetic disorder caused by mutations in a maternally inherited gene that is characterized by poor muscle control, limited speech, epilepsy and intellectual disabilities. Though there isn't a cure for the condition, new research at the UNC School of Medicine is setting the stage for one.

Unlike other single-gene disorders such as cystic fibrosis and sickle-cell anemia, Angelman syndrome has a unique genetic profile. Researchers have found that children with the conditions are missing the maternally inherited copy of the UBE3A gene, which regulates the levels of important proteins, while the paternally inherited copy of the UBE3A gene remains dormant. Missing a working copy of the gene leads to severe disruptions in brain development.

Ben Philpot, PhD

Hanna Vihma, PhD

But what if the dormant copy of the gene could be activated?

Ben Philpot, the Kenan Distinguished Professor of Cell Biology and Physiology at the UNC School of Medicine and associate director of the UNC Neuroscience Center, and his lab have identified a small molecule that could be safe, noninvasively delivered and capable of "turning on" the dormant paternally inherited gene copy brain-wide. A kind of gene therapy, this potential treatment could lead to proper protein and cell function for individuals with Angelman syndrome.

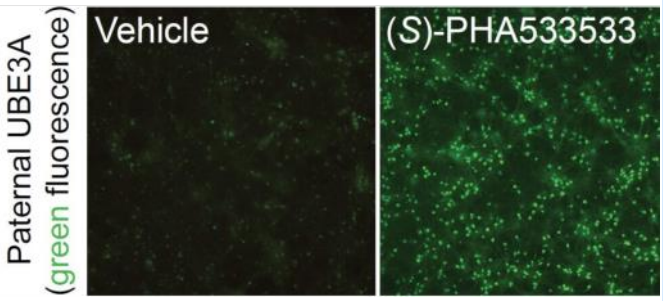
"This compound we identified has shown to have excellent uptake in the developing brains of animal models," said Philpot, a leading expert on Angelman syndrome and a member of UNC Lineberger Comprehensive Cancer Center. "We still have a lot of work to do before we could start a clinical trial, but this small molecule provides an excellent starting point for developing a safe and effective treatment for Angelman syndrome."

These results, published in *Nature Communications*, mark a major milestone in the field, according to Mark Zylka, the W.R. Kenan Jr. Distinguished Professor of Cell Biology and Physiology at the UNC School of Medicine and director of the UNC Neuroscience Center. No other small molecule compound has yet to show such promise for Angelman, he added.

Hanna Vihma, a postdoctoral research fellow in the Philpot lab and first author on the study, and colleagues screened more than 2,800 small molecules from a Pfizer chemogenetic library to determine if one could potentially turn on paternal UBE3A in mouse models with Angelman syndrome.

One compound, (S)-PHA533533, shows promise, but researchers are still working to identify the precise target inside cells that causes the desired effects of the drug. Philpot and colleagues also need to conduct further studies to refine the medicinal chemistry of the drug to ensure that the compound — or another version of it — is safe and effective for future use in the clinical setting.

"This is unlikely to be the exact compound we would take forward to the clinic," said Philpot. Along with medicinal chemists in the lab of Jeff Aubé, the Philpot lab is working to identify similar molecules with improved drug properties and safety profiles. "However, this gives us a compound that we can work with to create an even better compound that could be moved forward to the clinic."



Neurons show a bright fluorescent glow when treated with (S)-PHA533533, indicating that the small molecule potently activated the dormant paternal allele of UBE3A. Credit: Vihma et al 2024.

This work was supported by the Angelman Syndrome Foundation, the National Institute of Neurological Disorders and Stroke, the National Institute of Child Health and Human Development, the Simons Foundation, a sponsored research agreement between the University of North Carolina and Pfizer (to BDP), and Pinnacle Hill, LLC, a portfolio company of certain funds managed by Deerfield Management Company, L.P (to BDP).

NC-LEND 2024-2025 Trainees and Fellows

Ben Alschuler is a LEND Family Fellow, a Co-Chair of the Special Needs Advisory Council (SNAC) of Chapel Hill-Carrboro City Schools, and the parent of a child with autism. He is currently in the process of launching a non-profit organization, Hippo Campus of Chapel Hill, which will serve as a recreation center for neurodivergent youth and their families. Prior to becoming a neurodiversity advocate, Ben was a marketing executive who spent the past decade working to advance the growth of minority- and diverse-owned businesses through procurement opportunities and access-to-capital programs, including managing a $100M business lending program sponsored by Meta (Facebook) during the COVID-19 pandemic.

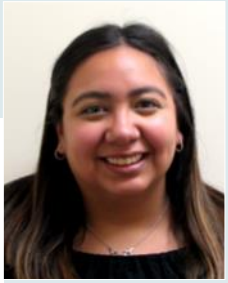
Eliah Anderson is a third-year doctoral student in the School Psychology PhD program at UNC-Chapel Hill. Eliah's professional interests focus on how psychological services can be expanded for youth and families living in rural, underserved communities.

Deja Barber is the Self Advocate Coordinator for HOPE NC. She has Cerebral Palsy and is actively involved in advocacy and disability work. She is currently working toward a double masters in School Counseling & Rehabilitation Counseling and is a graduate and teacher of the first Peer Mentor Certification for those with IDD in North Carolina. She is also a graduate and trainer of the Ability Leadership Project and has worked with the Disability Rights NC vaccine project. Deja aims to create a strong new generation of advocates who want to change the idea of disability.

Eyram Bossiade is a second-year master's student in the School of Social Work at UNC-Chapel Hill. Eyram's research and clinical interests include trauma-informed interdisciplinary behavioral health care, mindfulness-based self-compassion, disability justice advocacy, life course development, centering lived experiences, and promoting health and happiness.

Ciara Brown is a LEND fellow and a first year MPH student at UNC Gillings school of Public Health, concentrating on Nutrition and Dietetics (RD Training) at UNC Chapel Hill. Ciara hopes to serve as an advocate for children and adults with developmental disabilities in underserved communities throughout her journey as she starts her clinicals next summer.

Chelsea Calandra is a graduate student at UNC-Chapel Hill, working toward her degree as a Psychiatric Mental Health Nurse Practitioner, with plans to graduate in May 2025. She is committed to delivering equitable and holistic mental health care throughout all stages of life. Chelsea is particularly interested in neurodivergence and is dedicated to exploring the complexities of the human psyche to tailor her treatments to the unique needs of each individual.



Rosario Castillo is a predoctoral psychology intern working toward her PhD in School Psychology at UNC-Chapel Hill. Her work centers on supporting children and families, especially those facing educational and psychological challenges within the Latine community.

Emma Cochran is a Physical Therapist and the current Pediatric Physical Therapy Resident at UNC. Emma recently graduated with her Doctorate of Physical Therapy from High Point University, where she grew an interest in Pediatric PT, specifically working with children with Neurological Disabilities such as Cerebral Palsy, Traumatic Brain Injuries, Spinal Cord Injuries, Duchenne's Muscular Dystrophy, and other Neuromuscular conditions.

Faith Coleman is a 2nd-year Speech Language Pathology graduate student at North Carolina Central University. She is completing her Fall clinical practicum at UNC Hospitals pediatric specialty clinic for pediatric feeding disorders and hopes to specialize in pediatric feeding disorders after graduation. Faith completed her undergraduate degree at UNC-Chapel Hill in Psychology and History.

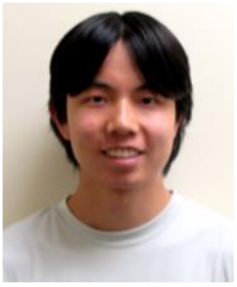
Kaitlin Cummings is a 4th year doctoral student in the UNC Dept of Clinical Psychology, working under the mentorship of Dr. Gabriel Dichter. She is passionate about translational research that informs the development of therapeutic interventions, and her current research focuses on the neurobiology underlying reward processing and immune functioning in autism spectrum disorder. In her clinical work, Kaitlin strives to support neurodiverse individuals and their families across the lifespan.

NC-LEND 2024-2025 Trainees and Fellows *continued*

Isabella Diaz is a master's student in Speech Language Pathology at UNC-Chapel Hill. Isabella's clinical interests include Pediatric Feeding Disorders, Early Intervention and Developmental Disabilities.

Sam Donnelly is a master's student at UNC and is a part of the Psychiatric and Mental Health Nurse Practitioner Program. Her research revolves around a Quality Improvement Project in the Emergency Department and its improvement of flow for people who enter the Emergency Department with psychiatric needs. Sam is interested in healthcare policy revolving around mental health and psychiatry in the state of North Carolina.

Penelope Franklin is a third-year student in the Clinical Doctorate of Audiology program at UNC. As a LEND trainee, she is interested in family-centered care and disability justice in the context of pediatrics. Her clinical and research interests involve intraoperative neuromonitoring and the reduction of care gaps between health care professionals and families.



Kevin Guo is an undergraduate at Duke University studying statistics and global health on the pre-MD/PhD track. He is interested in pediatrics and child health policy.

Emily Jedlowski is a third-year student in the Doctor of Audiology program at UNC-Chapel Hill. She graduated from Northwestern University with a Bachelor of Science in Human Communication Sciences. Emily's clinical interests include working with children with hearing loss and complex needs, equitable access to healthcare, and providing multidisciplinary patient-centered care. Her career aspirations are to work as a pediatric audiologist.

Mary Kathryn Phelps is a master's student in Speech Language Pathology at UNC-Chapel Hill. Mary Kathryn's clinical interests include functional therapy for children with intellectual and developmental disabilities, Augmentative and Assistive technology (AAC), and supporting independence and community engagement for adults with disabilities.

Chandler Knott is a doctoral intern in psychology. Chandler's research and clinical interests include early detection of autism in genetic syndromes characterized by intellectual disability, differential diagnosis of commonly co-occurring conditions in autism, parenting interventions for children with IDD, and access to school and community-based services and supports across the lifespan.

Erika Kristensen is in her second year of the Master of Science in Occupational Therapy program at UNC-Chapel Hill. As a former elementary school teacher, Erika is most interested in pediatric occupational therapy and loves working with kids. She is thrilled to have this opportunity with LEND to expand her knowledge, experience, and practice working with people in the IDD community and as an interdisciplinary team.

Johanna Lynch has worked with parents of children and young adults with disabilities for 15 years. She has represented parents on national- and state-level teams working to develop best practices in education and parent involvement to improve outcomes for students and their families.

Edith M. Nieves Lopez is a parent advocate of a neurodivergent child and board certified, bilingual pediatrician with 10+ years of experience working on the public health arena to bridge health disparities. Edith's special areas of interest include child development, neurodiversity, mental health, Latine experience and language justice. Edith is the founder of Developedia Consulting, an entity focused on healthy literacy to empower caregivers and institutions to serve marginalized communities in a way that addresses root causes of disparities.

NC-LEND 2024-2025 Trainees and Fellows *continued*

Talia Mango is a third-year Clinical Doctor of Audiology student at UNC. Her long-term goal within the field of pediatric audiology is to contribute to the development of techniques and practices used to diagnose hearing loss in children with severe and multiple disabilities and deafblindness.

Casey Martin is a third-year Doctor of Audiology student. Casey seeks to pursue a career in pediatric audiology to improve comprehensive care for children with hearing loss in addition to other developmental disabilities. She is a current member of the UNC Cochlear Implant Research Lab and is interested in exploring pediatric cochlear implant outcomes in children with complex needs.

Ali Marx is a research assistant at the UNC TEACCH Autism Program. Her research interests include adaptive functioning, quality of life, and community integration for adults with I/DD. She is passionate about connecting individuals with I/DD with services and resources in their community to help them live their best life. Her career goal is to become a Clinical Psychologist working with individuals with I/DD.

Madison McCall is a doctoral candidate in the clinical psychology program at UNC-Chapel Hill. Her research and clinical interests include disability-affirmative care, pediatric neuropsychology, and the development and implementation of digital psychological assessments that improve the accessibility and effectiveness of care delivery for children with developmental disabilities and chronic health conditions and their families. Formerly, Madison was a Health Policy Research Scholar at the Robert Wood Johnson Foundation and earned her B.S. in Biomedical Engineering at the University of Virginia.

Lindsay Mullin is a third-year student in the School Psychology PhD program. Lindsay's research interests include early detection and family-wide approaches to heterogeneity in neurodevelopmental disorders. Her clinical interests include providing fun and accessible care to children with neurodevelopmental disorders through assessment and individual therapy in school and community-clinic contexts.

Claudia Penny is a first-year student in the Genetic Counseling program at UNCG. Claudia's research interests revolve around mitochondrial DNA and the relationship between genotype mutations and phenotypic expression. She is clinically passionate about holistically teaching patients so they can make educated medical decisions for themselves.

Valentina Roa graduated from Wake Forest University. She is now in her fourth year of dental school at UNC Adams School of Dentistry. Valentina plans to become a pediatric dentist and hopes to use this LEND experience to become able and prepared to treat all children no matter their different abilities.

Sonny Russell is a second-year doctoral student in the Developmental Psychology program at UNC-Chapel Hill. Sonny's research interests include anti-ableism, supporting queer and gender-diverse autistic individuals, and equitable access to autism services across lifespan.

Isabella Russo is serving as the post-secondary education LEND trainee. She is currently in her final year of the MSW program. She is interested in accessible resources for the transition to adulthood for people with disabilities. This includes uplifting the voices of disabled individuals to ensure that they are being heard.

Dr. Lauren Schiff is a Minimally Invasive Gynecologic Surgeon in the UNC School Of Medicine's Department of OBGYN at UNC-Chapel Hill. She joined faculty in 2014 and since that time has held multiple operational and quality hospital leadership positions. She currently serves as the Associate Chief Medical Officer of Quality and Safety for UNC Medical Center. She is also actively engaged in patient care and teaching as a clinician and surgeon and is dedicated to caring for women with fibroids, endometriosis, complex benign gynecologic surgical conditions, and pelvic pain disorders. Her research has focused on clinical outcomes for care of surgical gynecologic disorders, the intersection of psychiatric mood disorder and gynecology care management and improving gynecologic care for autistic patients.

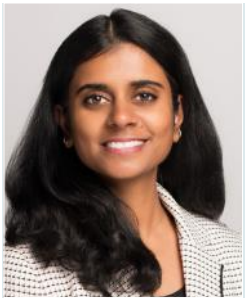
NC-LEND 2024-2025 Trainees and Fellows *continued*

McRae Scott is a graduate student in the Master of Social Work program at UNC-Chapel Hill. McRae's research and clinical interests focus on late diagnosis of autism spectrum disorder in adults assigned female at birth who exhibit high masking behaviors. They are also interested in adapting dialectical behavioral interventions for autistic individuals using the TEACCH framework, as well as exploring the complexities of differential diagnosis of borderline personality disorder and autism spectrum disorder.

Clara Thörn is a third-year doctorate student in the School Psychology PhD program at UNC-Chapel Hill. Her training has focused on the assessment and treatment of individuals with intellectual and developmental disabilities, with a particular focus on autism spectrum disorder (ASD). Clara's interests include early identification and intervention, as well as providing holistic support and care to individuals with ASD.

Allison Serdinsky is a self-advocate. She wants to learn how to change her future and make her life better through self-advocacy. She hopes that LEND will be a safe space to learn and grow.

Cheyanne Waller is a graduate student in the Audiology doctoral program. Cheyanne's clinical interests include early detection and diagnosis of hearing loss, bridging the gap between counseling and adherence to early intervention services, and parental engagement with early intervention services.



Suma Suswaram, PhD, a postdoctoral researcher at CIDD, conducts clinically relevant research to identify determinants of, and develop programs to enhance, communication skills in individuals with IDD. Her work applies socioecological and funds of knowledge theory and is rooted in cross-cultural and interdisciplinary principles.

Lauren Winfrey is a final-year Master's of Social Work student at UNC-Chapel Hill. Her clinical and research interests focus on individuals with I/DD and dual diagnoses. She is passionate about system enhancement and aims to better support individuals with disabilities and their families by improving access to inclusive resources.

Researchers Create New System to Decode Genetic Risk for Psychiatric Disorders

For many years, scientists have known that genetic variants, or differences in DNA code across people, play some role in neurological and psychiatric disorders. But the details were murky. Now, researchers at the UNC School of Medicine are using a combination of cell lines and DNA sequencing approaches to look closely at our genomes and identify which genetic variants and genes play roles in influencing one's risk for neurological and psychiatric disorders.

Jason Stein, PhD

A research team led by Jason Stein, PhD, associate professor of genetics and member of the UNC Neuroscience Center and the CIDD, has used a live-cell model system of the human brain to identify the function of genetic variants important for increasing the risk of developing schizophrenia, autism spectrum disorder, and bipolar disorder. The results were published in Nature Neuroscience.

"There are hundreds of different locations on our genome that are associated with psychiatric disorders," said Stein, who is also a member of UNC Lineberger Comprehensive Cancer Center. "But these locations are in regions of the genome where the function is not well understood. We supposed that some genetic variants function only when stimulated by certain neural pathways important for brain development."

Out of our entire genome, just 3% is responsible for creating codes that lead to the formation of proteins – the "machines" that perform needed tasks in our bodies. The other 97% of the genome does not code for proteins. It is in these "non-coding" regions where most genetic variants implicated in psychiatric illness can be found.

Non-coding variants are expected to be similar to light switches. They can "turn on" and "turn off" genes that code for proteins. But finding the precise function of these non-coding genetic variants has proven difficult for researchers. This is because "non-coding" genetic variants can have a "context dependent" function, which means they only work when specific cellular pathways are stimulated. In other words, the downstream effects of these genetic variants can only be observed when brain cells are alive and responding to stimulation.

The Stein lab decided to study the function of these genetic variants in neural progenitor cells, which are cells involved in brain development. Every cell line has a different genetic background, which allows researchers to compare and contrast genetic variants in both active and inactive states. Stein's lab members exposed the stem cells to different chemical compounds and controls to measure the differences in response.

These compounds stimulate the Wnt pathway, a cascade of proteins that play important roles in brain development. Using the living model, researchers found thousands of non-coding genetic variants that have a context-dependent function.

"Through the activation of Wnt-responsive genes, we found variants with context-dependent function that are implicated in schizophrenia risk," said Stein. "Finding these genetic variants represents an important step forward in our understanding of the mechanisms that cause someone to be at greater risk of developing a neuropsychiatric disorder."

Stein said that a similar study design using this live-cell model system of the human brain could be helpful for testing how genetic variation influences risk for environmental exposures, like lead exposure, and their impacts on the brain. Similarly, future applications of this approach could be used to prescribe psychiatric treatments based on an individual's genetics.

Co-first authors on the study were research associate Nana Matoba, post-doctoral fellow Brandon D. Le, and graduate student Jordan M. Valone.

Study Suggests Higher Amounts of Intervention May Not Be More Helpful for Children on the Autism Spectrum

When a child is diagnosed with autism, healthcare professionals often recommend intensive interventions, which can amount to 20-40 hours per week, to support their development. However, a study led by Micheal Sandbank, PhD, assistant professor in the Department of Health Sciences at the UNC School of Medicine and an IDDRC Investigator at the CIDD, and other researchers across the United States has found that more does not necessarily mean better.

Micheal Sandbank, PhD

Using data from 144 early childhood intervention studies, which involved 9,038 children between the ages of 0 to 8, researchers conducted a meta-analysis to determine whether higher intensity interventions provided increased benefits for young autistic children compared to less intensive interventions. They found that intervention outcomes did not improve as intervention intensity increased. Their results were published in JAMA Pediatrics. "We concluded that there was not rigorous evidence supporting the notion that increasing the amount of intervention produces better intervention outcomes," said Sandbank, who was first author on the study. "Instead, we recommend that practitioners consider what amount of intervention would be developmentally appropriate for the child and supportive to the family."

The most commonly recommended approach for autistic children in the United States is called "Early Intensive Behavioral Intervention," or EIBI. The current clinical guidelines regarding intensive intervention arose from a 1987 study which found that autistic children who received 40 hours of behavioral intervention per week had more cognitive improvement than those who received only 10 hours per week.

But many subsequent studies about behavioral intervention methods have provided mixed results and are lacking in quality. Of note, many studies have confused intervention amount with intervention approach, provided null results, or required retractions. In November 2023, Sandbank found that many low-quality studies are dominating the field and that few studies have adequately examined whether interventions can have adverse effects or harms. Notably, interventions requiring young children to be away from home for long durations of time can deprive them of critical rest, socialization with family members, and more. "In order to determine what amount of intervention is most effective, while also being minimally interruptive, we need more high-quality primary studies," said Sandbank. "Few high-quality studies systematically compare the same intervention offered at different amounts."

Many different types of intervention may be offered to young children on the autism spectrum. Behavioral interventions systematically teach functional and cognitive skills through direct one-on-one teaching and tend to be very intensive. Developmental interventions focus on improving children's engagement and social interaction through play with their caregivers and are frequently provided for only a few hours per week. Naturalistic developmental behavioral interventions blend behavioral and developmental approaches. All of these interventions can look very similar or very different in their implementation, depending on the provider.

In order to thoroughly investigate the impact of intervention amount, researchers measured it in three ways. They defined "intensity" as the amount of intervention provided within a given time frame (such as hours a day), "duration" as the total amount of time (in days) that intervention is provided, and "cumulative intensity" as an overall metric that describes the total intervention provided over the total duration. Using these three metrics, researchers explored whether intensity, duration, or cumulative intensity were associated with developmental benefit in young autistic children. At the same time, researchers wanted to determine if the strength of the relationship between the metrics and developmental improvement differed depending on the type of intervention provided. Their final sample for their meta-analysis included 144 separate studies involving a total of 9,038 participants. Knowing that neuroplasticity, or the brain's ability to adapt, is at its height during this developmental period and may affect the success of intervention, researchers controlled for participant age. They also accounted for the quality of included studies and intervention type with the help of meta-regression models.

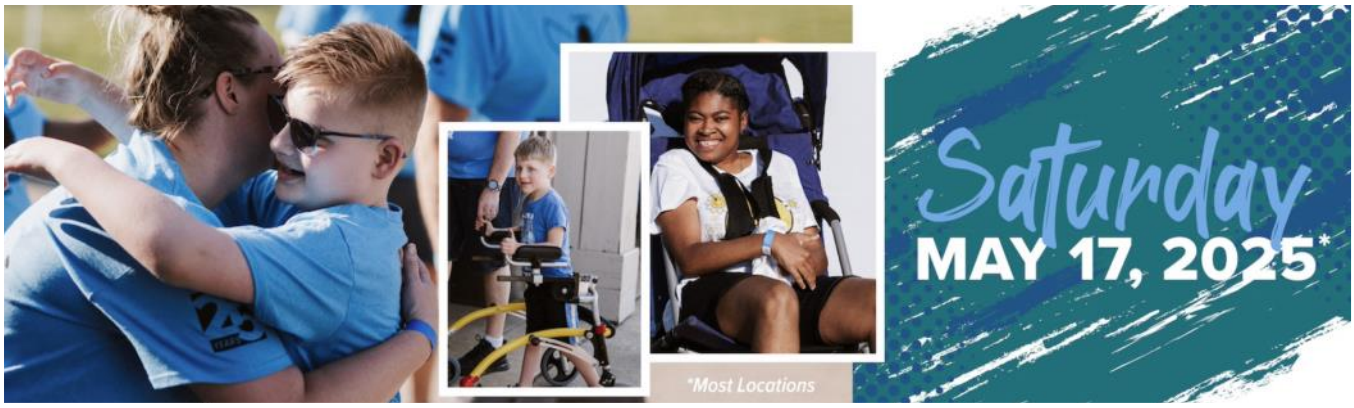
Taking all of these factors into account, researchers found no evidence that higher intensity interventions provided increased benefits for young autistic children. The evidence contrasts the results of quasi-experimental studies and some meta-analyses suggesting that high-intensity behavioral interventions are associated with more cognitive gains in young children on the autism spectrum. "There's probably a minimum amount of intervention needed to provide any benefit at all, and an optimal amount that is dependent on the child," said Sandbank. "Unfortunately, right now, we don't have clear evidence as to what that amount should be," said Sandbank.

This research suggests clinicians should avoid providing any specific amount of intervention as a default recommendation. Instead, clinicians should inform families that no single intervention amount is right for every child, and that a careful balance must be struck to meet the demands of intervention with other needs of the child to ensure that they thrive.

2024 Walk for Angelman Syndrome Foundation

Each year, the Angelman Syndrome Foundation (ASF) organizes nationwide fundraising walks to support families of children with Angelman Syndrome (AS). This year, members of the Clinical Trials Team co-coordinated the walk for North Carolina, which took place in Charlotte on May 18th. The walk brought together families from all over the state to build community and support their loved ones with AS. Several families that are involved in clinical trials and take advantage of other CIDD services were present at the walk. Many families look forward to this walk every year as a way to raise awareness and fundraise alongside the ASF. This year, the walk in North Carolina raised over $35,000 to fund critical research and trials, and support AS families. Many sites nationwide exceeded their fundraising goals. The walk is an inspiring and uplifting experience for everyone involved; the support for each other is tangible and serves as a reminder of why we do the work that we do. Thank you to everyone who helped fundraise over the course of this past year or otherwise showed their support. The team is excited to announce that we are hosting the 2025 walk in Chapel Hill.

Mark Shen, Caisi Hecht, and Hannah Rhiel enjoy building community with Angelman families at the 2024 NC Angelman Syndrome Foundation walk.



Support the Carolina Institute for Developmental Disabilities

The programs of the Carolina Institute for Developmental Disabilities provide innovative, high-quality clinical, research, and training activities supporting individuals with developmental disabilities. Now, more than ever, we need well-trained practitioners, teachers, and researchers. State funds pay only part of the costs to recruit and retain the best faculty and support the unique training and programs that are the hallmarks of the Carolina Institute for Developmental Disabilities experience. It is private funds that sustain and enhance these extraordinary opportunities for students, patients, families, and faculty. We can't do it without you!

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