CIDD Program Graduate Kenneth Kelty Wins National Honor

Kenneth Kelty, a graduate of the Leadership Education in Neurodevelopmental Disabilities Program at the Carolina Institute for Developmental Disabilities (CIDD), won The Tuesdays with Liz video contest, a national competition sponsored by the Association of University Centers on Disabilities (AUCD) to celebrate the 30th anniversary of Americans with Disabilities Act (ADA). Tuesdays with Liz is a weekly video series highlighting current issues in disability policy hosted by Liz Weintraub, a longtime disability advocate, and produced by AUCD.

LEND is funded by the Maternal and Child Health Bureau, a federal program within the Department of Health and Human Services, to support training of various health providers in the developmental disabilities field. The LEND program includes self-advocates who are individuals with developmental disabilities who themselves attend the program side by side with other individuals who do not have a developmental disability.

Mr. Kelty, who has autism, also won the Laura Lee Self-Advocate Leadership Award in 2017, the year he completed the LEND program. Thanks to the LEND Program at CIDD, Mr. Kelty said, “I was able to be a part of a wonderful leadership program for a person with a disability and for self-advocacy. And thanks to the ADA, I was able to get an education at an early age, which allowed me to have some inclusion with my typically-developing peers, and I was able to gain self-determination down the road.”

Mr. Kelty went on to graduate from Western Carolina, through its two-year University Participant Program. “Kenneth is a remarkable man,” said CIDD director Joseph Piven, MD. “He’s a tremendous advocate who often gives talks around the country about coping with developmental disabilities. He shows us every day how much we all can achieve.”

Mr. Kelty’s video can be viewed here: https://www.youtube.com/watch?v=MBsg8oz8Srs

Congratulations to Dr. Brittany Williams, Named as a Rising Star and Example of Black Excellence in Science

Brittany Williams, PhD, Postdoctoral Fellow in the T32 Postgraduate Research Training Program, has been named as a Rising Star on a list of inspiring black scientists compiled by Cell Press, a leading publisher of cutting-edge biomedical and physical science research and reviews.

Dr. Williams received her doctorate in Neuroscience under the tutelage of Dr. Amy Lee at the University of Iowa. Dr. Williams’ thesis work used electrophysiology and biochemical approaches to characterize the modulatory effects of visual disease causing mutations in voltage-gated Ca2+ channels. Now as a postdoctoral fellow at UNC, she works under the leadership of Dr. Ben Philpot, where she combines her expertise in electrophysiology and biochemistry with mouse models to understand how the overexpression of UBE3A impacts neurodevelopment and contributes to the manifestation of autism spectrum disorders (ASD).

Continued on page 4
UNC Intellectual and Developmental Disabilities Research Center (IDDRC)
Funded for Another Five Years

We are pleased to announce the successful competitive renewal of the UNC Intellectual and Developmental Disabilities Research Center (IDDRC), awarded by the National Institute of Child Health and Human Development (NICHD). The total award amount for this grant is over $1.24M per year for the next five years to support translational research on neurodevelopmental disorders, with the overarching goal of promoting research that leads to advances in prevention and treatment.

The UNC IDDRC is the primary research arm of the Carolina Institute for Developmental Disabilities (CIDD). Joe Piven, M.D., Director of the CIDD, Thomas E. Castelloe Distinguished Professor of Psychiatry and Pediatrics, has led the UNC IDDRC since 2000; Gabriel Dichter, Ph.D., Professor of Psychiatry, is Associate Director of the IDDRC.

One of 14 NICHD-funded IDDRCs around the US, the UNC IDDRC provides critical research, training and administrative support to 46 funded investigators, from multiple disciplines, spanning 13 departments throughout UNC, who are making significant contributions to understanding and treating neurodevelopmental disorders. While their expertise encompasses many of the currently recognized concentrations in IDD research, three particular strengths highlight the breadth, depth and clinical relevance of IDD research being conducted at UNC including: Early Development of Brain and Behavior, Autism and Related Syndromes; and, Early Detection and Intervention.

The Clinical Translational Core (CTC) includes the ‘Participant Registries’, directed by Gabriel Dichter, Ph.D. and co-director, Renee Clark, M.S.W., assisting investigators with recruitment of research subjects from four registries: the Autism Registry (6,699 subjects), the FX Registry (1,012 subjects), the General IDD Clinic Registry (225 subjects), and the Child Development Registry (13,236 subjects; 8,050 families); and, the Brain & Behavior Measurement Laboratories (BBML), led by Aysenil Belger, Ph.D., Professor of Psychiatry and Director of the Frank Porter Graham Child Development Institute, and Martin Styn, Ph.D., Associate Professor of Psychiatry and Computer Science, and providing support for characterizing brain and related metrics (e.g., novel software for processing infant brain images; support for infant/toddler EEG/ERP and eye tracking; consultation for MRI/DTI/fMRI studies) and consultation on behavioral assessment/study design/team development, led by Heather Hazlett, Ph.D., Assistant Professor of Psychiatry.

The Preclinical Core (PC), led by Sheryl Moy, Ph.D., Professor of Psychiatry and co-Director Ben Philpot, Ph.D. (Professor of Cell and Molecular Physiology and Associate Director, UNC Neuroscience Center), includes the Mouse Behavioral Phenotyping Laboratory, the Neuroscience Microscopy Facility, and the Small Animal Imaging Facility. Together, these components allow investigators to evaluate cell and animal-based models using a range of phenotyping approaches. The mouse behavior laboratory, led by Sheryl Moy, provides multi-domain phenotyping services for the characterization of novel genetic and environmental models of neurodevelopmental disorders, and has established optimal protocols for preclinical drug screens relevant to core ASD symptoms and other IDD abnormalities. The microscopy facility, directed by Michelle Itano, Ph.D., Assistant Professor and Director of the Neuroscience Center Microscopy Core (and recent recipient of a prestigious Chan Zuckerberg Initiative Grant to support innovative imaging science training), supports studies on cellular and molecular mechanisms in neurodevelopment, utilizing labeled neural tissue, brain slices, and intact, living embryonic brains. A recent expansion of this core, the Small Animal Imaging Facility, is led by Yen-Yu Ian Shih, Associate Professor of Neurology and Director of the UNC Center for Animal MRI. An overall goal for this core is integration linking abnormal behavioral phenotypes to changes in brain structure, connectivity, cellular function and molecular processes in animal models of human neurodevelopmental disorders.

Biomedical research is increasingly faced with the complex tasks of processing, analyzing, integrating and interpreting vast amounts of data from behavioral, imaging and genetic studies. The Data Science Core (DSC) therefore brings together expertise in biostatistics, psychometrics, bioinformatics, genetics/genomics and computer science to leverage overlapping resources and expertise required for state-of-the-art processing and analysis of biomedical data, and to expand opportunities for cross-disciplinary, collaborative research on IDDs. Directed by Yun Li, Ph.D., Associate Professor of Genetics (School of Medicine) and Biostatistics (School of Public Health), expert in statistical genetics, along with co-director, Young Truong, Ph.D., Professor of Biostatistics (School of Public Health), expert in biostatistics applied to image analysis, this core includes expertise in bioinformatics as applied to genetics and genomics research and biostatistics as applied to behavioral and neuroimaging studies. Additional support for this core comes from Guorong Wu, Ph.D., Assistant Professor of Psychiatry, providing expertise in computational biology; and, Sabrina Zadrozny, Ph.D., Director of the Data Management and Analysis Core at Frank Porter Graham Child Development Institute (FPG), providing support for statistical analysis of behavioral data with a particular emphasis on longitudinal studies and early development.

Since its inception in 1967, the UNC IDDRC has had a major impact on developmental disabilities research and scientific training at UNC. With this successful NICHD renewal, we will be able to continue to pursue the overarching goal of promoting research that leads to important advances in the prevention and treatment of IDDs.
HEELS Prep, an interdisciplinary collaboration designed to support young adults with intellectual disabilities, concluded seven weeks of online programming in response to the COVID-19 pandemic. Through weekly virtual group classes and individual coaching sessions, HEELS Prep addressed key areas of the transition to adulthood, including self-management, life skills, career development, mental health, and community safety. It offered diversified learning experiences and practical application of the rights, responsibilities, and benefits of adulthood.

Dara Chan, program director, noted the significant role of graduate students to the program's success as it moved online from in-person programming. "We have an incredible team, and in this year’s summer program our graduate students really shined," Chan said. “They took the lead with support and direction from our instructors and staff to quickly transform our program. As coaches, they provided valuable support to our participants to make what they were learning in their classes more meaningful in their day-to-day lives.”

Jacklyn Googins, co-coordinator of HEELS Prep, a Leadership Education in Neurodevelopmental Disorders (LEND) trainee, and a second-year master's student studying occupational therapy, said program participants showcased resilience and adaptability. “It was uncharted territory, but it also opened up new modalities for communication. All of the participants developed the technical skills needed to meaningfully access the Zoom platform and express their ideas in a variety of ways.”

Adela Van Name, the project manager and mother of a HEELS Prep participant, said bringing the program’s lessons into the home environment via video allowed for continued support from family members. “The families were gracious in letting us come in,” Van Name said. Depending on participants’ goals, tasks often involved cleaning, organizing, grooming, and cooking.

Julie Doran, a second-year student in the Division of Clinical Rehabilitation and Mental Health Counseling, said each participant worked one on one with a goals and career coach. “The concepts really come to life when it’s translated to their everyday experience,” Doran said. An interdisciplinary team of students and staff met weekly with each other to offer varying perspectives on program initiatives. This also allowed for individualized modifications for the lessons and identifying gaps in service. This year’s 10 participants included six program alumni from the inaugural year. Van Name said both returning and new parents were impressed by the participants’ increased sense of purpose and self-determination.

Googins said HEELS Prep encourages a self-directed model of learning, which emphasizes the ability of adults to function well when they direct their life, their goals, and have support. “The program involved the participants teaching each other and, in many cases, the instructors learning from them,” she said. At the conclusion of the program, participants self-reported increased feelings of confidence in each of the key areas of adulthood. Greg Boheler, also a LEND trainee and second-year master’s student in occupational therapy, said the coaching component allowed for greater individualization based on personal interests, preferences, and aspirations for adult life.

The HEELS Prep team is exploring a fall 2020 program in order to ensure opportunities for continued learning and support for program participants. HEELS Prep is part of the HEELS 2 Transition organization, an interdisciplinary collaboration among the UNC School of Medicine’s Department of Allied Health Sciences, the Carolina Institute for Developmental Disabilities, UNC TEACCH Autism Program, and community partner Kidpower, Teenpower, Fullpower, International.
Congratulations to Dr. Brittany Williams, Named as a Rising Star and Example of Black Excellence in Science continued

The goal of Dr. Williams’ research is to not only better understand (mechanistically) how such disorders arise, but to identify novel therapeutic strategies to correct and/or prevent the manifestation of ASD symptoms associated with the overexpression of UBE3A. View the lists from Cell Press:

100 Inspiring Black Scientists in America: http://crosstalk.cell.com/blog/100-inspiring-black-scientists-in-america
100 More Inspiring Black Scientists in America: http://crosstalk.cell.com/blog/100-more-inspiring-black-scientists-in-america

In May of this year, Dr. Williams was also awarded a Burroughs Wellcome Fund Postdoctoral Enrichment Program Fellowship. The Postdoctoral Enrichment Program (PDEP) provides a total of $60,000 over three years to support the career development activities for underrepresented minority postdoctoral fellows in a degree-granting institution in the United States or Canada whose training and professional development are guided by mentors committed to helping them advance to careers in biomedical research.

“Diversity drives discovery,” said Fund President and CEO Dr. Louis Muglia. “Through fostering innovation and creativity and providing a strong mentoring element this program enables recipients to become part of a strong and growing network of postdoctoral researchers who are making successful transitions to independent careers.”

New Grant to the Intellectual and Developmental Disabilities (IDDRC) Network: Harnessing Clinical Genomic Characterization to Accelerate Translational Advances for Patients with IDD

Over the last decade there has been unprecedented progress in understanding the genetic susceptibility to IDD. Rare mutations (copy number and sequence variants) are now being identified in ~ 30% of individuals with intellectual and developmental disabilities who undergo clinical genomic testing (e.g., chromosomal microarray or whole exome sequencing). While identification of these genomic characteristics is getting more and more common, our understanding of how to link these genomic variants to specific clinical features in an individual patient have lagged.

In this joint effort across the network of 14 NICHD Intellectual and Developmental Disabilities Research Centers (IDDRCs), together with their respective CTSA’s (NIH funded ‘Clinical Translational Service Awards’), this project aims to more fully and systematically characterize genes relevant to intellectual and developmental disabilities. The ultimate goal of this effort is to (1) develop and implement a systematic strategy to characterize clinical characteristics relevant to IDDs that can be used across the field; (2) identify relevant genes and their specific relationship to clinical features (so-called ‘curation’); and, (3) establish a national, CTSA-IDD patient registry across the country of all the information compiled and develop a platform to be able to link this information to patient clinical data in the electronic health records (EHRs) in health care systems around the country. UNC is one of the three lead sites along with Washington University in St Louis and Harvard University. The UNC site is led by Heather Hazlett, PhD, and Joe Piven, MD. This work builds on the pioneering efforts of the NIH-funded, ClinGen Initiative, where UNC has had a longstanding leadership role under the direction of Jonathan Berg, MD, PhD, a Professor in the Department of Genetics at UNC and an IDDRC investigator.

Laura Hiruma Selected as a Children's Mental Health Champion

Laura Hiruma, PhD, psychologist and clinical assistant professor at the CIDD, has been selected to serve as a Children's Mental Health Champion for the 2020-2021 cycle. The award is funded by the Association of University Centers on Disabilities (AUCD) and supported by a cooperative agreement from the Centers for Disease Control and Prevention (CDC). Children's Mental Health Champions serve as state liaisons to the CDC’s Children's Mental Health program and positive parenting resources; act as community champion change agents to increase awareness activities and improvement of positive parenting practices; develop and test strategies for building partnerships to promote identification, screening, referral, and intervention; and implement effective strategies for mental health promotion, prevention supports, and creation of networks (e.g., school, healthcare, and community) and better connections between these systems.
New Faces at the CIDD

Jamie K. Capal, MD, is an Associate Professor with dual appointments in the CIDD and Neurology. She completed her residency in Pediatrics, Neurology with special qualification in Child Neurology, and Neurodevelopmental Disabilities at Cincinnati Children’s Hospital Medical Center in 2012. Her clinical and research interests involve individuals with neurogenetic conditions that result in neurodevelopmental disabilities. Dr. Capal was recruited to the CIDD to help lead clinical trials and translational research in neurodevelopmental conditions, including Angelman syndrome, Tuberous Sclerosis Complex, and others neurogenetic conditions. She will also have a clinical presence at the CIDD.

Jessica Girault, PhD, has joined the faculty at the CIDD as a Research Assistant Professor with a dual appointment in the Department of Psychiatry. She earned her PhD in Neuroscience from UNC in 2018 and completed her postdoctoral training under the mentorship of Dr. Piven as a T32 Fellow at the CIDD. Dr. Girault’s program of research incorporates pediatric neuroimaging, clinical assessments, and molecular and behavioral genetics to study early brain development in neurodevelopmental disorders. Currently, Dr. Girault is the principal investigator of projects funded by the National Institutes of Mental Health and the Foundation of Hope to study how genetic background and genetic risk factors shape brain and behavioral development in autism spectrum disorder, Fragile X syndrome, and Down syndrome.

Rebecca Grzadzinski, PhD, has joined the faculty at the CIDD as a Research Assistant Professor with a dual appointment in the Department of Psychiatry. She earned her PhD in Clinical Psychology from Teachers College, Columbia University in 2018 and completed her postdoctoral training under the mentorship of Drs. Heather Hazlett and Linda Watson as a T32 Fellow at the CIDD. Dr. Grzadzinski’s program of research examines early child and family characteristics that impact functional outcomes in children with neurodevelopmental disorders (e.g., autism spectrum disorder; ASD), behavioral and symptom change over time (e.g., treatment response), and associations with brain markers. Currently, Dr. Grzadzinski is the principal investigator of a project funded by the National Institutes of Mental Health incorporating behavioral phenotyping with neuroimaging to study social communication skills, and brain correlates, in children with Down Syndrome and/or ASD.

Ben Philpot Receives Awarded from the Simons Foundation Autism Research Initiative

Seizures are an extreme outcome of hyperexcitability in the brain and are a common neurologic complication in autism spectrum disorder (ASD), with estimates ranging from 5-40% of ASD individuals also suffering from seizures. Seizures are even more common in syndromic forms of ASD. For example, almost all (>90%) individuals with Angelman syndrome suffer from seizures, in addition to severe intellectual disability and lack of speech. In this grant funded by the Simons Foundation Autism Research Initiative (SFARI), the Philpot lab will study the mechanisms that cause seizures in Angelman syndrome, with the expectation that this knowledge will also guide therapeutically oriented investigations for seizures and more mild manifestations of brain hyperexcitability across the broader spectrum of autism.
Jack Roush Receives CIDD Impact Award

Professor Jackson Roush, Division of Speech and Hearing Sciences, received the 2019 CIDD Impact Award from the Carolina Institute for Developmental Disabilities. Established in 2016, the CIDD Impact Award recognizes the outstanding and lasting contributions of a professional, family member or self-advocate within the NC developmental disability community, toward improving the quality of life for individuals and their families. The award was presented on December 29th by Dr. Joe Piven, Director of CIDD, in recognition of Dr. Roush’s many years of service to CIDD and to the North Carolina LEND program. His most recent roles include P.I. for the NC-LEND Program and Project Director for the LEND Pediatric Audiology Grant. Dr. Roush was also instrumental in establishing CIDD’s Hearing and Development Clinic which provides interdisciplinary assessments for children who are deaf or hard of hearing with autism or other co-occurring conditions.

UNC NeuroSpark Research Award Winners Celebrated

Four research teams at the UNC School of Medicine won $25,000 pilot grants with the goal of increasing collaborations throughout the neurosciences, as part of a UNC NeuroSpark event.

As part of the Forward Together Strategic Plan, the UNC School of Medicine committed $100,000 this year to fund pilot neuroscience research projects. The goal of this funding is to increase collaborations in the neurosciences and to incentivize faculty to generate pilot data for use in grants to the NIH and other external agencies. This is the first year of a five-year commitment for NeuroSpark from the school of medicine.

Eight neuroscience research teams at the school of medicine competed for four $25,000 pilot grants. Each team gave a 12-minute pitch to the audience. Following the presentations, the audience voted for the top four projects. The four teams that received the most votes were selected as winners.

The event was attended by more than 100 graduate students, postdocs, lab members, and faculty.

Congratulations to the winning teams, which include three teams led by IDDRC Investigators:

“A Pilot PET/MR Study of Neuroinflammation in Autism”
Gabriel Dichter, PhD, associate professor in the UNC Department of Psychiatry; Director, Clinical Affective Neuroscience Lab
David Lalush, PhD, associate professor in the UNC/NCSU Joint Department of Biomedical Engineering
Erin Walsh, PhD, assistant professor in the UNC Department of Psychiatry

“Targeting Sleep Disruption in the Progression of Alzheimer’s Disease”
Graham Diering, PhD, assistant professor in the UNC Department of Cell Biology and Physiology
Todd Cohen, PhD, assistant professor in the UNC Department of Neurology

“Building a Resource of Genetically Diverse Human Post-Mortem Brain Tissue”
Jason Stein, PhD, assistant professor in the UNC Department of Genetics
Hyejung Won, PhD, assistant professor in the UNC Department of Genetics
Leigh Thorne, MD, professor in the UNC Department of Pathology and Laboratory Medicine
Worldwide Study Unlocks Genetic Secrets of Gray Matter

A major international collaboration co-led by UNC School of Medicine scientist Jason Stein, PhD, has produced the first genetic map of the cerebral cortex, identifying more than 300 genetic variants that influence cortical structure and play roles in psychiatric conditions.

The cerebral cortex is the relatively thin, folded, outer “gray matter” layer of the brain crucial for thinking, information processing, memory, and attention. Not much has been revealed about the genetic underpinnings that influence the size of the cortex’s surface area and its thickness, both of which have previously been linked to various psychiatric traits, including schizophrenia, bipolar disorder, depression, attention deficit hyperactivity disorder (ADHD), and autism.

Now, for the first time, over 360 scientists from 184 different institutions – including UNC-Chapel Hill – have contributed to a global effort to find more than 200 regions of the genome and more than 300 specific genetic variations that affect the structure of the cerebral cortex and likely play important roles in psychiatric and neurological conditions.

The study, published in Science, was led by co-senior authors Jason Stein, PhD, IDDRC investigator and assistant professor in the Department of Genetics at the UNC School of Medicine; Sarah Medland, PhD, senior research fellow at the QIMR Berghofer Medical Research Institute in Australia; and Paul Thompson, PhD, associate director of the Mark and Mary Stevens Neuroimaging and Informatics Institute at the University of Southern California. Ten years ago, these scientists cofounded the ENIGMA Consortium, an international research network that has brought together hundreds of imaging genomics researchers to understand brain structure, function, and disease based on brain imaging and genetic data.

“This study was only possible due to a huge scientific collaboration of more than 60 sites involved in MRI scanning and genotyping participants,” Stein said. “This study is the crown jewel of the ENIGMA Consortium, so far.”

The researchers studied MRI scans and DNA from more than 50,000 people to identify 306 genetic variants that influence brain structure in order to shed light on how genetics contribute to differences in the cerebral cortex of individuals. Genetic variants or variations are simply the slight genetic differences that make us unique. Generally speaking, some variants contribute to differences such as hair color or blood type. Some are involved in diseases. Most of the millions of genetic variants, though, have no known significance. This is why pinpointing genetic variants associated with cortex size and structure is a big deal. Stein and colleagues consider their new genetic roadmap of the brain a sort of “Rosetta stone” that will help translate how some genes impact physical brain structure and neurological consequences for individuals.

Among the findings of the research published in Science:

• Some genetic variants are associated with cortical folding, measured as surface area, while other genetic variants are associated with the thickness of the cortex.

• Genes that determine surface area are related to very early development in the fetal cortex, while thickness appears to be driven by genes active in the adult cortex.

• People at genetic risk for depression or insomnia are genetically inclined toward having lower surface area, while people with a genetic risk for Parkinson’s disease tend to have higher surface area.

• The vast scale of the project allowed the discovery of specific genes that drive brain development and aging in people worldwide.

“Most of our previous understanding of genes affecting the brain are from model systems, like mice,” Stein said. “With mice, we can find genes, knock out genes, or over express genes to see how they influence the structure or function of the brain. But there are a couple of problems with this.”

One problem is, quite simply, a mouse is not a human. There are many human-specific features that scientists can only study in the human brain.

“The genetic basis for a mouse is very different than the genetic basis for humans,” Stein said, “especially in the noncoding regions of the genome.”

Continued on next page
North Carolina’s Early Check Program to Add New Muscular Dystrophy Test to Newborn Screening Panel

The test will identify Duchenne muscular dystrophy and some other types of muscle conditions, as part of the Early Check Program, which is led at UNC-Chapel Hill by Cynthia Powell, MD.

RTI International, a nonprofit research institute, is partnering with the Muscular Dystrophy Association and Sarepta Therapeutics to add a test for Duchenne muscular dystrophy and some other types of muscle conditions to the Early Check program’s screening panel for North Carolina newborns.

Early Check is an ongoing partnership between RTI, Duke University, UNC-Chapel Hill, Wake Forest University, and the North Carolina State Laboratory of Public Health that offers free, voluntary tests for conditions not included in the state’s standard newborn screening. The program, which currently screens for fragile X syndrome and spinal muscular atrophy, is offered to all new parents in North Carolina. The UNC site principal investigator is Cynthia Powell, MD, Professor of Pediatrics and Genetics at the UNC School of Medicine, Director of the Medical Genetics Residency Program, member of the UNC Children’s Research Institute and the Intellectual and Developmental Disabilities Research Center, and CIDD clinician in the Angelman Syndrome clinic.

“Adding this test to Early Check with the help of the Muscular Dystrophy Association and Sarepta is a significant step forward for parents and newborns in North Carolina,” said Holly Peay, PhD, a senior research public health analyst at RTI. “Early detection of conditions like Duchenne muscular dystrophy is critically important because it can allow for earlier treatment for the baby and education and counseling for the family.”

Duchenne muscular dystrophy is a genetic disorder characterized by progressive muscle degeneration and weakness that can be life-limiting. About 1 in 3,500-5,000 boys are born with DMD, and very rarely girls can also be affected. Past research has shown that the average age of diagnosis for Duchenne muscular dystrophy is 4- to 5-years old, long after symptoms first appear.

“Sarepta is pleased to partner on the Duchenne Early Check program. This is an important initiative in our broader commitment to ensure that families have equitable access to early diagnosis and the best chance for improved outcomes as a result of earlier care and treatment,” said Diane Berry, PhD, Senior Vice President, Global Policy, Government & Patient Affairs at Sarepta.

“We are committed to promoting policies and programs that move newborn screening forward for neuromuscular disorders,” says Lynn O’Connor Vos, president and CEO of MDA. “We are delighted to be working with RTI and its partners to bring free newborn screening for DMD to families in North Carolina.”

The new screening test, which earned FDA approval in December of 2019, is expected to be added to the Early Check panel in late spring or early summer of this year. If infants are confirmed to have Duchenne muscular dystrophy or a similar condition, Early Check will provide families and clinicians with tailored educational materials, genetic counseling, and targeted consultation and assessment through Duke University. The Early Check program will collaborate with other ongoing pilots towards a shared goal of understanding the potential benefit of newborn screening for Duchenne.

For more information about Early Check, visit: www.earlycheck.org or the Muscular Dystrophy Association, visit: www.mda.org.
Welcome T32 Postdoctoral Trainees

The CIDD T32 Postgraduate Research Training Program develops researchers with expertise in both the biological basis and clinical manifestations of neurodevelopmental disorders. This broad-based and integrated perspective enables researchers to better relate across disciplines and maximizes the potential for major research advances in understanding the pathogenesis and treatment of these disorders.

Kelly Caravella, PhD
Mentors: Heather Hazlett, PhD; Karen Grewen, PhD

Dr. Kelly Caravella earned her doctorate in Clinical-Community Psychology from the University of South Carolina under the mentorship of Dr. Jane Roberts. Dr. Caravella’s research focuses on the longitudinal development of autism symptomatology in infants and toddlers with fragile X syndrome (FXS). As a T32 postdoctoral fellow, Dr. Caravella will be working under the mentorship of Drs. Hazlett and Grewen to compare neurobiological and behavioral markers of autism spectrum disorder across groups of infants at high risk for developing autism spectrum disorder (ASD), specifically, infants with FXS, infant siblings of children with ASD and infants who experienced prenatal drug and alcohol exposures. Dr. Caravella's program of research aims to identify risk and protective factors in these high risks groups to inform early detection and intervention efforts to improve quality of life and support optimal developmental outcomes.

Dea Garic, PhD
Mentors: Joseph Piven, MD; Mark Shen, PhD

Dr. Dea Garic received her doctorate in psychology from Florida International University, with a dual concentration in developmental science and cognitive neuroscience. Under the mentorship of Dr. Anthony Dick, Dea utilized diffusion weighted imaging (DWI) methods to assess microstructural properties that might underlie typical and atypical brain development and how early structural connectivity relates to language and executive function behavioral outcomes in young children. As a postdoctoral fellow at UNC, Dea will be applying her DWI expertise to collaborative investigations examining how axonal properties and cerebrospinal fluid (CSF) flow contribute to neurodevelopmental disorders. The overarching goal of this work is to potentially identify non-invasive biomarkers of symptomatic progression and may provide avenues for targeted, biologically-based interventions for children with Angelman Syndrome and autism spectrum disorder.

Meredith Gruhn, PhD
Mentor: Margaret Sheridan, PhD

Dr. Meredith Gruhn received her doctorate in clinical psychology from Vanderbilt University under the mentorship of Dr. Bruce Compas. A central theme in Dr. Gruhn’s research involves the neurodevelopmental and psychosocial consequences of early life adversity (ELA). As a T32 fellow under the mentorship of Dr. Margaret Sheridan, Dr. Gruhn will examine (a) neural, physiologic, and cognitive-behavioral sequelae of adverse experiences and (b) etiologic and exacerbating roles of ELA in the presentation of neurodevelopmental disorders (e.g., ADHD). The long-term goal of this work is to inform, develop, and disseminate interventions for high-risk youth.
Celebrating Two Retirements at the CIDD

Congratulations to Teresa McCrimmon and Deb Zuver
Thank You for Your Many Years of Dedicated Service to the University!

Teresa McCrimmon retired at the end of July from the CIDD with 30 years of state service. We were lucky enough to get Ms. McCrimmon from P&A where she had not only learned the systems, but also developed insurance and billing skills. At the CIDD Ms. McCrimmon managed and coordinated the clinical billing and insurance while providing expertise and leadership to the entire team. She led more than one successful system transition with the type of knowledge that can only be obtained from a long successful career. Ms. McCrimmon helped the many clinics and clinicians at CIDD, always with a smile, and her expertise and she will be sorely missed. Her colleagues said, “She always took the time to help regardless of how busy she was.” Another colleague noted that “Miss T was clever when it came to insurance companies and how to best work within the ever changing system.” The Deputy Director, Jeffry Low, said, “I will miss her as a wonderful person and a fantastic resource for the Institute. She is a special lady and we were all fortunate to call her a friend and co-worker.” Thank you, Ms. McCrimmon! We are grateful for the years of service you gave to CIDD and to others with whom you worked.

Deborah “Deb” Zuver retired from the CIDD at the end of April and she is already missed! Ms. Zuver had multiple roles at the CIDD through the years, most often focused on advocacy and inclusion. This was evidenced by her leadership in Project STIR and, as LEND faculty, Ms. Zuver helped identify and strengthen the role of the self-advocates in the cohort of trainees each year. Additionally, she assisted the LEND program in developing a LEND faculty slot for a self-advocate who, in turn, provides critical coaching and mentoring for LEND trainees each year. As one colleague noted, “Ms. Zuver not only brought a smile to work each day, but brought skills and expertise in empowering self-advocates to reach their goals.” Ms. Zuver helped coordinate and host the statewide Inclusive Postsecondary Education (IPSE) Alliance that meets quarterly to better understand and share resources about educational opportunities in NC. She spearheaded an initiative to establish an IPSE program (HEELS UP) at UNC-CH and, following a successful IPSE Summit in 2019 involving faculty, administrators, families and IPSE graduates from other programs, a pilot program was approved at UNC-CH and is expected for Summer 2021. Ms. Zuver has served on many CIDD and statewide committees focused on advocacy, guardianship, sexual violence, etc. One co-worker noted, “Not only does Ms. Zuver have a wealth of knowledge and experience regarding I/DD, but she also connects with people in a way that truly values and amplifies their voices. In addition, her warmth and authenticity fostered a collaborative environment at CIDD.”

Another colleague described Ms. Zuver this way, “Ms. Zuver exemplifies how to advocate for people with disabilities and differences by meeting every person with respect and expectation, expectation that each individual has something valuable to contribute and deserves an opportunity to do so. She doesn’t talk inclusion; she lives it.”

2020 CIDD “VIRTUAL” Community Talk Series Presents:

Law Enforcement and ID/DD: Strategies for Success

Sara E. Stanton, MA, NCC - Clinical Director – NC START East

This FREE presentation will explore the interactions between law enforcement and individuals with intellectual and developmental disabilities as well as provide strategies for increasing successful outcomes integrating a systems perspective

WHEN: Wednesday, September 9, 2020 from 6:30PM to 8:00PM
WHERE: Register here: http://tinyurl.com/y6537l6x

At the time of registration, you will receive a Zoom link to join the Community Talk Series.

Please direct any questions to Debbie Reinhartsen, Ph.D., CCC-SLP at Debbie.Reinhartsen@cidd.unc.edu
LEND Trainee Spring Research Symposium 2020

The 2019 – 2020 LEND cohort completed a successful training year in April despite challenges resulting from COVID-19. The LEND program quickly moved to an online format for remaining classes and for the 2020 Research Symposium. As part of their LEND program, all trainees are expected to complete a research project and present it to the group at the conclusion of the year. The trainees did an exceptional job of developing and pre-recording their presentations and being available for questions from the audience. 25 projects were presented over two 3-hour virtual sessions and topics were varied and of high interest. Examples of projects and presentations ranged from Nutrition Issues in Children with Spina Bifida to Promoting Visibility of Inclusive Post-Secondary Opportunities in North Carolina. Attendance for the virtual event was impressive and appreciated as all trainees and numerous CIDD faculty and mentors participated in both sessions. The table highlights each trainee’s presentation:

Dan Earixson (Post-Secondary)
Promoting Visibility of Inclusive Post-Secondary Opportunities in North Carolina
Dan’s project described the collaborative process used to organize the Virtual College Fair for students with I/DD, an event aimed at promoting awareness of inclusive post-secondary education opportunities. The inclusive post-secondary alliance visibility team reached networks and agencies across the state to promote this event and reach the intended audience. Roadblocks to implementation and next steps in response to the current pandemic were discussed, as well as the results from the team’s outreach.

CJ Hacker (Occupational Therapy)
The Sky's the Limit: Air Travel and the I/DD Community Through an Occupational Lens (A Research-Based Toolkit)
CJ created a multifaceted, research-based toolkit to address three barriers to air travel for individuals with I/DD. The toolkit includes resources and recommendations compiled using the unique clinical reasoning of an occupational therapist. In keeping with OT’s frequently occupied role as a connector, this toolkit is designed to be of use to distinct yet undeniably intertwined populations: individuals with I/DD and their families, airline professionals, and students/practitioners who desire to learn about and work to close the gaps herein.

Mara Lewis (Self-Advocate)
LGBTQIA+ Community: What it’s Like Being an Ally
Mara’s project was about the LGBTQIA+ Community, Equality and being a Straight Ally. Members of the LGBTQIA+ Community are more likely to have a disability than the general population. Mara defined the community terms, discussed being an ally, gave personal examples, and included websites and tips for how to get involved.

Betty Martinez (Nursing)
Asthma in Children with Intellectual and Developmental Disabilities: Tools for Exceptional Kids with Asthma (TEKA) Kit
Betty presented an asthma tool kit that will provide children and families asthma information tailored for children with IDD and asthma. The objective of her project was to pilot and analyze the qualitative responses on the use, perception of the tool, and the nature of having a child with the comorbid condition of asthma; with a follow up plan to share and educate colleagues at the military clinic about communication strategies they can use in practice for families with children who have co-occurring asthma and IDD with a goal to improve patient outcomes.

Molly Marus (Social Work)
Assessment of the Healthcare Needs of Older Autistic Adults
Molly’s project focused on the healthcare service needs of autistic adults over the age of 49. She examined data from a pilot study on aging and autism conducted at the CIDD between 2011-2014. Molly looked at the reported healthcare needs for physician services, nursing services, allied health services (not including mental health), and mental health services, using the NC-SNAP measure. The presentation covered background information on the need for such information, findings, discussion, project limitations, and directions for future research.
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| **Johnna Miller (Physical Therapy Fellow)**  
*Effectiveness of guided discussion with parents of pediatric physical therapy clients in improving patient outcomes.*  
Johnna’s project focused on enhancing the family-centered approach while treating pediatric patients and their families. She presented a case series in which she incorporated guided discussion into her weekly treatment sessions. The goal of this case series was to determine if an increase in parental satisfaction will improve patient/family compliance and patient outcomes with physical therapy. |
| **Caroline Garrett (Social Work)**  
*Investigating Outcomes and Experiences of Females on the Autism Spectrum*  
Many people with Autism Spectrum Disorder, particularly women, may learn to camouflage or mask their autism characteristics. Without a diagnosis, women do not have access to appropriate services, a community of similar individuals, or an explanation for their differences. Through an anonymous survey with 87 respondents, Caroline analyzed age of diagnosis, social masking traits, mental health outcomes, quality of life, and more to provide information about this often-overlooked group of people with developmental disabilities. |
| **Meredith Pysnik (Family Fellow)**  
*Caring for the Caregiver: Supporting Self-Compassion in Parents of Children with Disabilities*  
Meredith’s LEND project focused on addressing a challenge experienced by most—if not all—parents of children with disabilities: managing care for themselves in the midst of managing all of the other overwhelming details of daily life. Meredith developed an accessible series of guidelines and practices for parents of all backgrounds to use in beginning work toward self-compassion, and companion series of guidelines and practices for providers to use as they work to support self-compassion in parents that they encounter. |
| **Rose Valentine (Genetic Counseling)**  
*Atypical Deletions in Angelman Syndrome*  
Rose’s LEND project is an ongoing literature and case history review to document similar phenotypic presentations in patients with atypically large deletions and Angelman syndrome. Rose reviewed the clinical presentation and mechanisms of Angelman syndrome, preliminary findings from the literature review and cases seen at the CIDD, and the importance of characterizing and expanding the phenotype of Angelman syndrome to include these atypical deletion patients. |
| **Rachel Sandercock (Clinical Psychology)**  
*Dyadic Vocal Contingency in Infants at High Risk for Autism Spectrum Disorder*  
Identifying the earliest emerging signs of ASD is a priority for understanding how the disorder develops and unfolds, as well as for shaping and facilitating the provision of early intervention services. Rachel presented a study that will examine contingency between infant and adult vocalizations utilizing a high-risk infant sibling sample recruited through a larger prospective longitudinal study (the Infant Brain Imaging Study Network). The goal of this study is to examine contingency as a potential mechanism underlying prodromal differences in social-communicative behaviors by quantifying the extent to which it mediates the relationship between early attention and later diagnostic and language outcomes. |
| **Brittany Brisson (Counseling)**  
*Resource Navigation for Persons with Intellectual and Developmental Disabilities*  
Brittany collaborated with families to create a website that makes resources easy to search for, find, and access. The resulting website also allows individuals and families to comment on their experiences with hospitals and residential facilities through a moderated comment feature. The site includes general search features as well as search by broader categories. |
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<td>Ashley White (Nutrition)</td>
<td>Nutrition Issues in Children with Spina Bifida Myelomeningocele</td>
<td>Ashley’s LEND project was a clinical case study completed in partnership with the dietitians from the pediatric feeding team at UNC Children’s Hospital. Ashley observed a 15-month-old female with spina bifida myelomeningocele (SBM) for her case during a routine monthly check-up visit at the pediatric clinic. Ashley’s presentation covered the background information about SBM, the current nutrition recommendations for children with SBM, and a brief synopsis of the clinical case she observed at UNC Children’s Hospital.</td>
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<td>Brittany Anderson (Social Work)</td>
<td>Police Involvement for Individuals with I/DD Experiencing a Crisis</td>
<td>Brittany examined rates of police involvement for individuals with I/DD who experienced a crisis. She analyzed crisis call data from NC START between July 2018 and February 2020. Brittany looked at correlations between race, gender, and county of residence. A chi-squared test was performed on each set of data to see if there were statistically significant connections between these demographic characteristics and police response.</td>
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<td>Madeleine Barclay (Audiology)</td>
<td>Use of CDC’s Learn the Signs. Act Early. (LTSAE) Materials in NC’s Early Hearing Detection and Intervention (EHDI) Program</td>
<td>The Centers for Disease Control and Prevention (CDC) initiated the Learn the Signs. Act Early. (LTSAE) program to engage parents and other early care providers in developmental monitoring aimed at facilitating earlier identification of children with developmental delays and disabilities. Madeline’s presentation: 1) examined conditions that co-occur in children who are deaf or hard of hearing, 2) described LTSAE materials developed by the CDC to promote early identification of developmental delays and disabilities in young children, and 3) described how the CIDD and NC-LEND are collaborating with the North Carolina EHDI program to promote and integrate LTSAE resources in our state.</td>
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<td>Katie Bishop (Social Work)</td>
<td>An Evaluation of the Dialectical Behavioral Therapy Chill Skills Group</td>
<td>Katie evaluated the progress of clients participating in the Chill Skills Dialectical Behavioral Therapy Group at the Carolina Institute for Developmental Disabilities. This is a therapy group for individuals with co-occurring intellectual and developmental disabilities and mental health diagnoses. Data was collected through the Life Problems Inventory, which measures client’s levels of emotional dysregulation, confusion of self, interpersonal chaos, and impulsivity.</td>
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<td>Hannah Silverstein (Maternal Child Health)</td>
<td>Using Raven’s Distractors to Estimate Reasoning Skills among Malawi Youth</td>
<td>In Western country settings, Raven’s Progressive Picture Matrices are traditionally used to measure non-academic cognitive reasoning, relying on images as opposed to language or math. Hannah’s research is a secondary data analysis from an intervention study in Malawi that included eight Raven’s items on the baseline questionnaire for 2,514 youth ages 13-19. Her goal was to estimate cognitive scores using this short set of items that was normalized for this population. Hannah explored three different ways of calculating cognitive ability: sum scores, two parameter logistic item response theory (IRT), and nominal response model IRT.</td>
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<td>Chelsea Carter (Speech/Language)</td>
<td>Effects of Professional Development in Teaching Writing for Students with Disabilities</td>
<td>Chelsea’s project was a systematic review to investigate the impact of professional development in the area of writing for teachers on academic outcomes for students with intellectual, developmental or learning disabilities. This systematic review is part of a larger meta-analysis from the UNC Writing Skills Development Program on the effects of professional development in writing on improving student writing ability across a range of studies. The authors conducted the initial search and de-duplication followed by screening for inclusion (title/abstract and full text), quality appraisal, and data extraction. The limited results showed the positive impact of professional development for teachers and students.</td>
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<td>William Crosby (Nursing)</td>
<td>Development of a Low-Cost Speech-Generating Device</td>
<td>The aim of William’s project was to develop a low-cost speech generating augmentative and alternative communication (AAC) system. William sought to reach this goal through adapting commercial off the shelf technology that would generate voice output, therefore enhancing low-tech communication boards. He first developed a proof of concept using the PENpal optical identification reader system and Core Board. Success of the proof of concept led to further refinement resulting in a first prototype that is scalable, durable, provides voice output in multiple languages, and costs less than $150.</td>
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<td>Baraka Harris (NC Central)</td>
<td>Speech and Language Therapy in Fragile X Syndrome: Information Sheet</td>
<td>Baraka created a resource for speech-language pathologists to use to guide treatment decisions for children with Fragile X Syndrome (FXS). The resulting information sheet contains the appropriate goal areas, intervention programs and treatment strategies that are recommended in research literature and treatment guidelines for children with FXS. Multiple resources, including videos, websites and references to guidelines and peer-reviewed articles, are included on the document for clinicians to access.</td>
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<td>Karen Hall (Education)</td>
<td>Self-Identified Strengths for Children (SIS-C)- A Semi-Structured Clinical Interview for Children</td>
<td>Karen presented the Self-Identified Strengths for Children (SIS-C), a semi-structured clinical interview designed to be used with children from ages 3 – 21 years with a variety of abilities. It is an interview given to a child that gathers information about the child’s strengths in a variety of settings. Several adaptations and modified protocols are also presented to support various levels of communication as well as functional abilities. The SIS-C was developed to increase a child’s voice in the evaluation process and to provide IEP teams with valuable information about a child’s self-identified strengths.</td>
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<td>Camille Herring (Title V/Act Early)</td>
<td>Reconsideration of Race, Ethnicity, and Culture in Early Identification and Treatment of Autism in Black Children</td>
<td>Camille’s presentation was a systematic review of barriers that African American families face to receive autism diagnostic and intervention services for their children. Along with the systematic review, her presentation also examined the research currently being done to inform next steps that should be considered when studying race and autism. Rather than just using race as the main determinant of health, Camille’s presentation also explored other considerations such as honing in on different cultures within African Americans.</td>
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<td>Felicia Williams (Family Fellow)</td>
<td>IDD Services Checklist</td>
<td>Felicia created the intellectual and developmental disabilities (IDD) services checklist with the goal of helping the community track services and organize plans for individuals with IDD based on their age. The ages are grouped based on services criteria and marked transition of life stages. This way, no matter where individuals and families are on the journey, they can quickly connect with what is most important to them at any given time. Ultimately, this one page, front and back document allows easy mobility for everyone and may be reformatted to the preference of services organizations, providers and families.</td>
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<td>Crisma Emmanuel (Nursing)</td>
<td>A Meta-Synthesis of the Experiences of People with ASD, and that of their Family Members, in Acute Care and Inpatient Settings</td>
<td>Crisma completed a systematic review that included only qualitative articles published within the last ten years and focused on the experiences of individuals with autism, and their family members, in acute care and inpatient settings. Nine articles were included, with six total themes being identified. Themes were separated into two categories: Challenges and Barriers and Identified Suggestions and Necessary Improvements. Overall, more research is necessary, with an emphasis on collecting more firsthand accounts from individuals with ASD about their experiences in acute care settings.</td>
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Meredith Braza, Jenna Duerr, Courtney Schlachter, Amy Spicer (Audiology)
Cochlear Implantation in Children with Neurodevelopmental and Related Disabilities

Meredith, Jenna, Courtney and Amy presented a study that retrospectively probes the outcomes of 97 cochlear implant users with one or more developmental disabilities receiving services from the Children’s Cochlear Implant Center at The University of North Carolina at Chapel Hill. Number of comorbidities and cochlear nerve status were both statistically significant variables in cochlear implant success, and children were more likely to receive one cochlear implant as opposed to two. Notable was the number of syndromes and comorbidities in this sample, with 35 conditions total represented.

LEND Trainee and Third-Year Doctor of Audiology Student Meredith Braza Receives Scholarship from American Academy of Audiology Foundation

The American Academy of Audiology Foundation has selected third-year Doctor of Audiology student Meredith Braza as a recipient of its Continued Achievement Scholarship. The selection process includes a review of the nominee's academic record, letters of recommendation, and professional aspirations which in Meredith’s case include a special interest in working with children facing multiple challenges.

Meredith was one of only a few students selected in her first year for participation in the NC-LEND program. In her second year she participated in another federally funded pediatric audiology grant (U.S. Department of Education) with a focus on developmental disabilities in children who are deaf or hard of hearing. Meredith also has a special interest in research. Over the summer of 2020 she was selected for a prestigious NIH-T35 summer research experience at Boys Town National Research Hospital.

As she begins her third year, Meredith is working as a research assistant in UNC’s Speech Perception and Auditory Research Lab in the Department of Otolaryngology. LEND faculty member, Dr. Jack Roush, Professor in the Division of Speech and Hearing Sciences, describes Meredith as “a motivated and multi-talented young woman who combines technical ability, interpersonal skill, genuine passion for working with children and families, and an extraordinary willingness to go above and beyond.”

LEND Audiology Trainees Receive Best Poster Award at Annual Early Hearing Detection and Intervention Conference

Four NC LEND students from UNC’s Doctor of Audiology (AuD) program received the award for ‘best poster’ at the national 2020 Early Hearing Detection and Intervention (EHDI) conference in Kansas City, Missouri, held in early March. The ongoing project, which is focused on improving hearing screening in the neonatal intensive care unit (NICU), is a collaboration between NC-LEND and the North Carolina EHDI program in the Division of Public Health. The LEND audiology trainees include (L-R) Katie Sobon, Stephanie Ortega, Shannon Muzio, and Jane Khin with UNC faculty mentor Dr. Jackson Roush and Dr. Marcia Fort, Unit Manager for Genetics and Newborn Screening, NC Division of Public Health.
Developmental Origins of Cortical Surface Area Hyperexpansion in Autism

Rose Glass, neuroscience doctoral student in the lab of Jason Stein, PhD, IDDRC investigator and Assistant Professor of Genetics, and faculty in the UNC Neuroscience Center, is pictured at the November 2019 IDDRC Director’s meeting with Joe Piven, MD, presenting about developing induced pluripotent stem cells from individuals at high risk for autism. This research is funded by NICHD and the Foundation of Hope.

Your Support

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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