



Inside the Institute

Issue # 17
September 2015

Newsletter for the Carolina Institute for Developmental Disabilities

UNC Scientists Pinpoint How a Single Genetic Mutation Causes Autism

The research shows the precise cellular mechanisms that lead to the disorder and how an existing drug might help thousands of people with autism.

Last December, researchers identified more than 1,000 gene mutations in individuals with autism, but how these mutations increased risk for autism was unclear. Now, UNC School of Medicine researchers are the first to show how one of these mutations disables a molecular switch in one of these genes and causes autism.

Published online August 6, 2015 in the journal *Cell*, the research shows that an enzyme called UBE3A can be switched off when a phosphate molecule is tacked onto UBE3A. In neurons and during normal brain development, this switch can be turned off and on, leading to tight regulation of UBE3A. But a research team led by Mark Zylka, PhD, Associate Professor of Cell Biology and Physiology, found that an autism-linked mutation destroys this regulatory switch. Destruction of the switch creates an enzyme that cannot be turned off. As a result, UBE3A becomes hyperactive and drives abnormal brain development and autism.

"Genetic studies are showing that there will be about 1,000 genes linked to autism. This means you could mutate any one of them and get the disorder. We found how one of these mutations works," said Zylka, senior author of the *Cell* paper and member of the UNC Neuroscience Center and the Carolina Institute for Developmental Disabilities (CIDD).

The work was done in human cell lines, as well as mouse models. Because this one autism-linked UBE3A mutation was part of the Simons Simplex Collection – and Zylka previously had been funded through a Simons Foundation grant – he had access to the cells that were used to find this one mutation. When Jason Yi, PhD, a postdoctoral fellow in Zylka's lab and former CIDD T32 postdoctoral trainee, sequenced the genes from the cell samples – including cells from the child's parents – he found that the parents had no hyperactive UBE3A but the child did.

The child's regulatory switch was broken, causing UBE3A to be perpetually switched on. "When this child's mutation was introduced into an animal model, we saw all these dendritic spines form on the neurons," said Zylka. "We thought this was a big deal because too many dendritic spines have been linked to autism."

Their findings thus pointed to hyperactivation of UBE3A as the likely cause of this child's autism. It was previously thought that too much UBE3A might cause autism because duplication of the 15q chromosome region – which encompasses UBE3A and several other genes – is one of the most commonly seen genetic alterations in people with autism. This is called Dup15q Syndrome.

As part of their study, Zylka and Yi found that protein kinase A (PKA) is the enzyme that tacks the phosphate group onto UBE3A. This finding has therapeutic implications, particularly since drugs exist to control PKA.

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Mark Zylka, PhD



Jason Yi, PhD
(Photos by Max Englund,
UNC Health Care)

Capitol Hill Briefing on Mysteries of the Brain: Frontiers in Neuroscience

Spencer Smith spoke on Capitol Hill about how new technologies and multidisciplinary research are advancing our understanding of the brain. At this congressional briefing organized by the House Appropriations Subcommittee on Commerce, Justice and Science and National Science Foundation on Thursday, July 9th, the panelists discussed new technologies and recent discoveries pertaining to the Brain Research through Advancing Innovative Neurotechnologies (BRAIN) Initiative.

Spencer Smith (pictured far right) shaking hands with Congressman Chaka Fattah; Professor Gary Lynch, University of California, Irvine (pictured left); Professor Scott Thompson, University of Maryland (middle).



Neurogenetic Developmental Disorders Clinic to Open This Fall

We are pleased to announce that our Neurogenetic Developmental Disorders Clinic will be opening in the fall 2015. The Neurogenetic Developmental Disorders Clinic is an interdisciplinary evaluation service for children, adolescents, and adults with genetic syndromes or documented chromosomal abnormalities associated with developmental disability (e.g., Fragile X Syndrome, Williams Syndrome, Down Syndrome, 22q11 Deletion Syndrome, and others). Patients with [Angelman Syndrome](#) and [Prader Willi Syndrome](#) will most often be served by our other clinics dedicated to those particular disorders.



Pictured left to right: Drs. Debbie Reinhartsen, Laura Politte, Emily Kertcher, Heather Hazlett

The clinical team -- comprised of Laura Politte, M.D., Child and Adolescent Psychiatry; Heather Hazlett, Ph.D., Psychology; Emily Kertcher, Ph.D., MOTR/L, Occupational Therapy; Debbie Reinhartsen, Ph.D., CCC-SLP, Speech and Language Pathology; (additional disciplines will be determined as needed) -- will provide assessment of cognitive strengths and weaknesses, neuropsychiatric symptoms, fine motor skills, sensory processing differences, and speech/ language concerns that may be part of a genetic disorder. Ongoing monitoring and consultation will be available for clinic patients if needed. Referral to geneticists, genetic counselors, educational consultants, and other medical specialists may be initiated as needed. At the end of the consultation session, a report will be provided along with assistance in identifying local resources.

For more information about the Neurogenetic Developmental Disorders Clinic contact Twyla Peoples at 919-843-1529.

UNC Scientists Pinpoint How a Single Genetic Mutation Causes Autism *continued from page 1*

"We think it may be possible to tamp down UBE3A in Dup15q patients to restore normal levels of enzyme activity in the brain," Zylka said. "In fact, we tested known compounds and showed that two of them substantially reduced UBE3A activity in neurons."

One of the drugs, rolipram, previously had been tested in clinical trials to treat depression but was discontinued due to side effects. One of the symptoms associated with Dup15q syndrome is sudden unexpected death in epilepsy. In light of these life-threatening seizures, Zylka pointed out that it may be worth examining whether lower doses of rolipram, or other drugs that increase PKA activity, provide some symptom relief in Dup15q individuals. "The benefits might outweigh the risks," he said.

The research was recently featured on WRAL. Click on the link below to watch video.

[UNC Researchers
Unlocking the Mysteries of Autism](#)

<http://www.wral.com/lifestyles/healthteam/video/14815615/>



CIDD Partners with State Agencies to Implement the National Core Indicators

The CIDD has worked closely with the Division of Mental Health, Developmental Disabilities and Substance Abuse Services since 1999 to implement the National Core Indicators (NCI) project. NCI is a voluntary effort by state developmental disability agencies to gauge their own performance using a common and nationally validated set of measures. These measures allow states to evaluate and improve services for individuals with developmental disabilities and their families. Through comparison of North Carolina's performance to that of other states, as well as comparing current and trend data within NC, statewide policies and procedures can be evaluated and improved. Additionally, by examining NC's performance over time, the state can monitor how it has improved and determine what remains to be done.

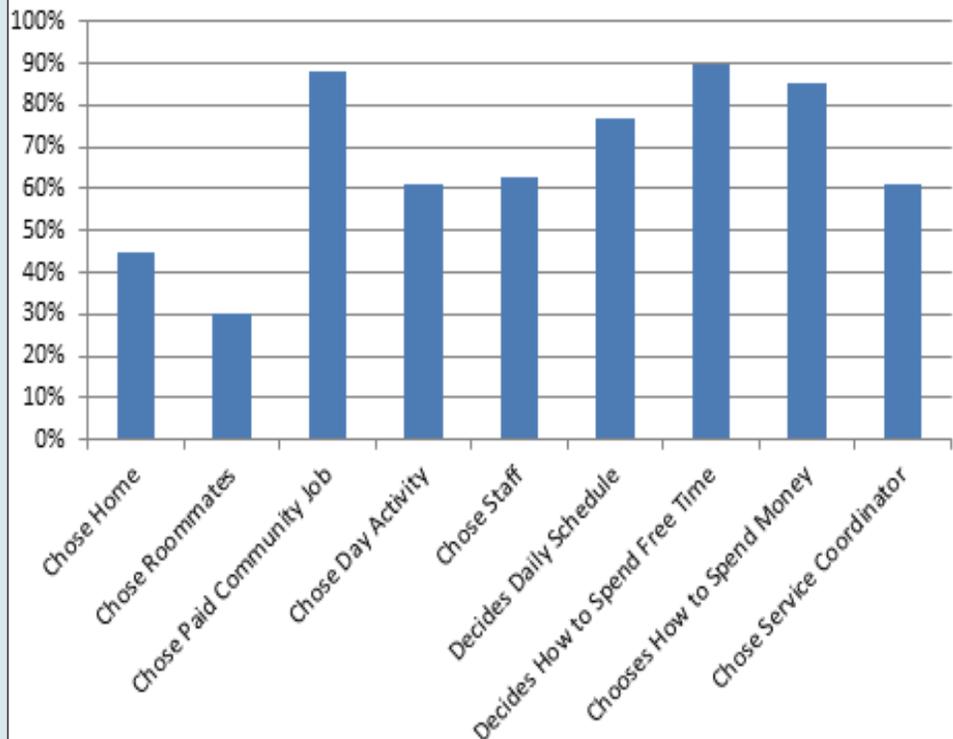
NCI has developed a set of more than 100 standard performance measures (or "indicators") that states use to assess the outcomes of services provided to individuals and their families.

These indicators focus on areas such as: employment, rights, service planning, community inclusion, choice, health, and safety.

CIDD implements two sources of NCI data collection: family surveys (e.g., satisfaction with supports) and consumer surveys (e.g., empowerment and choice issues). Three different surveys are mailed yearly to a random sample of families with a child receiving services, families/guardians of an adult who lives at home, and families/guardians of an adult who lives away from home.

The primary source of data collection is the NCI Adult Consumer Survey, an interview conducted with a random sample of individuals age 18 or older who are receiving at least one publicly funded service.

**NCI 2013-2014 North Carolina
Individual Outcomes: Choice and Decision Making**



CIDD interviewers meet with individuals to ask questions about where they live and work, the kinds of choices they make, the activities they participate in within their communities, their relationships with friends and family, and their health and well-being. CIDD conducts interviews with approximately 750 – 800 individuals per year and provides this information to the DMHDDSAS.

Dr. Karen Feasel heads this quality improvement project for NC and says, "Hearing directly from individuals and their families allows the Division to develop a fuller picture, one that incorporates people's own experiences and perceptions, of service outcomes across the state. Our longstanding partnership and the professionalism and skill of CIDD's project staff and field interviewers have proven over the years to be instrumental to the success of the NCI project in North Carolina."

For more information and past reports: <http://www.nationalcoreindicators.org>

CIDD is also pleased to have partnered this year with the NC Division of Aging and Adult Services to pilot the NCI-Aging and Disabilities (NCI-AD) Adult Consumer Survey. NCI-AD is an initiative designed to support states' interest in assessing the performance of their programs and delivery systems and improving services for older adults, individuals with physical disabilities, and caregivers. The primary aim is to collect and maintain valid and reliable data that give states a broad view of how publicly-funded services impact the quality of life and outcomes of service recipients.

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CIDD Welcomes Visitors Andy Imparato and Sharon Lewis

The CIDD was pleased to have a visit in May from Andy Imparato, JD, Executive Director of the Association of University Centers on Disabilities (AUCD). AUCD is a membership organization that supports and promotes a national network of university-based interdisciplinary programs including UCEDDs, LENDs and IDDRCs. CIDD is one of only 8 states that have all 3 of these federal disability programs at one site. Mr. Imparato met with Joe Piven and Jack Roush, directors of our UCEDD, LEND and IDDRC, and was given an overview of all three programs. In addition, he talked throughout the day with faculty and trainees. Discussion centered on current and future training initiatives, CIDD activities that promote the Developmental Disabilities Act, and future directions within the field of intellectual and developmental disabilities.



Pictured left to right: Jack Roush, Ann Palmer, Jean Mankowski, Deb Zuver, Joe Piven, Andy Imparato, Becky Pretzel, Donny Yerby, Debbie Reinhartsen



CIDD also welcomed Sharon Lewis in May; she serves in the dual roles of Principal Deputy Administrator of the US Administration for Community Living, as well as HHS Secretary Burwell's Senior Advisor on Disability. Ms. Lewis has an outstanding reputation for her work in disability policy and advocacy issues, including postsecondary education (PSE) opportunities. She met with Donna Yerby and Deb Zuver to discuss their important efforts in PSE and the advancements in North Carolina.

CIDD Partners with State Agencies to Implement the National Core Indicators *continued from page 3*

NCI-AD brings an important value proposition to the field of aging and disability services through knowledge development on indicators and outcomes that assess quality of life, community integration, and person-centered services. This knowledge development will help to address long-recognized gaps in measuring indicators and outcomes in LTSS service systems that go beyond measures of health and safety to address important social, community, and person-centered goals.

For more information: <http://www.nasud.org/initiatives/national-core-indicators-aging-and-disabilities>.

NCI efforts at CIDD are led by Becky Pretzel and Debbie Reinhartsen, with invaluable support from Rebecca Greenleaf, Thomas Castelloe, Deborah Zuver and our interviewers across the state.

Innovative Gene Transfer-Based Treatment Approach

UNC School of Medicine researchers have developed an innovative, experimental gene transfer-based treatment for children with giant axonal neuropathy (GAN).

Researchers led by Steven J. Gray, PhD, Assistant Professor in the Department of Ophthalmology and a researcher in UNC's Gene Therapy Center and Carolina Institute for Developmental Disabilities, developed the experimental treatment in studies conducted at UNC. Gray's work in this area was funded almost entirely by Hannah's Hope Fund, a charity founded by the parents of Hannah Sames, an 11-year-old girl with giant axonal neuropathy (GAN), to support the development of a treatment and cure. This extremely rare genetic disorder causes children to gradually lose the ability to balance themselves, move their muscles and to feel certain sensations. Most children born with GAN do not survive beyond their early 20s because of progressive impairment of their ability to breathe.



Steven J. Gray, PhD
(photo by Max Englund/UNC Health Care)

The treatment approach developed at UNC uses a genetically modified virus called AAV to deliver a missing gene, the gigaxonin gene (scAAVg/JeT-GAN), into the cerebrospinal fluid of children with GAN. The therapeutic viral vector to be used in each of these injections is prepared at the UNC Vector Core Human Applications Laboratory. A clinical trial of this approach is now underway at the National Institute of Neurological Disorders and Stroke (NINDS) of the National Institutes of Health (NIH) in Bethesda, Maryland. The first patient was enrolled in May. This is the first gene delivery approach directly into the spinal fluid in order to treat an inherited neurological disorder, and is expected to pave the way to developing treatments for many other related diseases.

Gray chose to focus his career on this rare genetic condition after meeting Hannah, who is the same age as his own daughter, Aubrey.

"This has been a coordinated and committed effort between Hannah's Hope and UNC to drive a treatment forward for GAN. Hannah's Hope is a truly amazing community that provides a constant source of inspiration. Our goal has always been to bring hope to the families affected by this devastating disease, and we are proud to be taking the first step to making a GAN treatment a reality," said Dr. Gray. "We are greatly appreciative of NIH/NINDS for partnering with us on this life-saving mission. This trial is the first in history to deliver gene therapy through the spinal fluid to test the potential to achieve broad treatment of the spinal cord and brain (central nervous system or CNS). It is a momentous step forward, and we're already seeing clear application of this approach to treat other diseases studied in my lab."

Gray serves as an Associate Investigator on the trial as does R. Jude Samulski, PhD, director of the UNC Gene Therapy Center. "After 30 years of focusing on optimizing successful gene delivery, it is very rewarding to finally see these approaches being tested for some of the unmet clinical needs caused by these terminal genetic disorders," Samulski said. "This specific study represents a culmination of years of basic research from the UNC Gene Therapy Center and that primarily of Steve Gray's team coupled with clinical expertise at the NIH. More importantly, this journey for me has personally been a truly rewarding one that started seven years ago with a parent knocking on the office door asking if we could 'help save her child', to last week's gene therapy administration; a remarkable and humbling journey that I'm privileged to be a part of."

Carsten Bönnemann, MD, who is leading the trial at NIH said, "This first intrathecal (into the spinal fluid) delivery of a viral gene therapy vector in a human patient is a fundamental step towards developing a causal treatment for giant axonal neuropathy (GAN), a devastating progressive neurogenetic disorder of childhood. At the same time it is also paving the way for similar gene transfer based treatments for many other neurological disorders in which nerve cells of the spinal cord and brain need to be targeted, including spinal muscular atrophy. Bringing such path-breaking treatments to children affected by neurogenetic disorders is really the core mission of our team here at the NINDS so we are very excited to be helping to move this approach to a clinical trial. That this first step is now being taken is testament to Hannah's Hope Fund and Dr. Steve Gray's tenacity and enormous commitment, but also to the courage of our first young patient who volunteered to receive this treatment, and others who will follow."

The Phase I clinical trial, which officially started in January, seeks to enroll a total of up to 20 patients with GAN who are ages five and older (ClinicalTrials.gov Identifier: NCT02362438). Each of the children and their families will be required to live within 100 miles of the NIH for two months after receiving the gene transfer treatment, which will be given by a single injection by spinal tap into their cerebrospinal fluid, which flows around the brain and spinal cord.

Spencer Smith Receives 2015 McKnight Technological Innovations in Neuroscience Award



Congratulations to CIDD Investigator, Spencer Smith, PhD, recipient of a 2015 McKnight Technological Innovations in Neuroscience Award for his research in multiphoton imaging for large brain volumes. The McKnight Technological Innovations in Neuroscience Awards support scientists working on new and unusual approaches to understanding brain function. The program seeks to advance and enlarge the range of technologies available to the neurosciences.

Unlike many other types of cells, neurons have a remarkable degree of autonomy. Individual neurons act together in complex ways to shape thoughts and behaviors, but it is tremendously difficult to study this process because we lack appropriate tools to observe these interactions. Multiphoton imaging, which can resolve individual neurons from millimeters away, appears to be the path forward. In previous research with two-photon microscopy, Dr. Smith's lab developed optical technology to view individual neurons firing in the mouse brain over an area of 1.5 square millimeters; his team then expanded the range to a span of 9.6 square millimeters. Now he is seeking to build a custom optical system to gain access to 1 million neurons (about one-fifth of the neurons in the mouse neocortex) while retaining high resolution to observe neurons individually. By making it possible to image neural activity across large brain volumes, the technology has significant potential to advance neuroscience research. Read more about the [McKnight Technological Innovations in Neuroscience Awards](#).

Mark Zylka Awarded Rett Syndrome Research Trust Grant for Long Genes Screening



Mark Zylka, PhD, Associate Professor in the Neuroscience Center and the Department of Cell Biology and Physiology at UNC, discovered that a class of drugs called topoisomerase inhibitors reduces the expression of long genes, raising the possibility that this class of drugs could be clinically relevant for Rett. One such drug is topotecan which is FDA approved for cancer. While the lab of Michael Greenberg at Harvard Medical School is currently testing topotecan in Rett mice models, the drug may not be ideal since it doesn't get into the brain easily and would be toxic for long term use. As a result, Dr. Zylka has been awarded \$400,000 from Rett Syndrome Research Trust (RSRT) to screen for other compounds that can rebalance expression of long genes safely. For more information visit the [RSRT website](#).

Jeffrey Stirman Awarded Burroughs Wellcome Fund Grant



Jeffrey Stirman, Ph.D., has been awarded a grant of \$500,000 as part of the Burroughs Wellcome Fund's Career Awards at the Scientific Interface (CASI) program. This award recognizes his excellent and important technical contributions to biomedical science, and will support his early career.

Dr. Stirman's early training was in physics, and he published several first-authors papers in the field. He later earned a PhD in Bioengineering, where he created sophisticated optical devices for patterned optogenetic stimulation of freely moving animals. Most recently, he developed new imaging technology to measure neural activity in different parts of the brain simultaneously, with single neuron resolution. He is using the system to better understand function of neural circuitry, and what might be disrupted in complex neurological diseases including autism. Dr. Stirman is a postdoc in Spencer Smith's lab, which is part of the Department of Cell Biology and Physiology, the Neuroscience Center, and the Carolina Institute for Developmental Disabilities. To read the announcement, visit the [Burroughs Wellcome Fund website](#).

Garret Stuber Receives Hettleman Prize



Garret Stuber, Ph.D., Assistant Professor of Psychiatry, Cell Biology and Physiology and member of the UNC Neuroscience Center and the Carolina Institute for Developmental Disabilities, has been awarded the highly prestigious 2014 Philip and Ruth Hettleman Prize for Artistic and Scholarly Achievement.

Dr. Stuber, who joined the Carolina faculty in 2010, has achieved an international reputation for his work on brain circuitry associated with adaptive and maladaptive behaviors. Through his groundbreaking research in dissecting brain reward circuits, Dr. Stuber and his colleagues have pinpointed the cellular connections that can trigger addictive behaviors, aversive behaviors and obesity. This understanding could lead to the development of ways to regulate the activity of cells in a specific region of the brain, and ultimately to new treatments for eating disorders.

"He has found very surprising and important new organizational principles that underlie complex motivated behaviors," said William Snider, director of the UNC Neuroscience Center. "Dr. Stuber's work is highly multidisciplinary. It spans genetic and molecular biological approaches, advanced physiological techniques for monitoring neural activity in living mice, and behavior analysis."

Dr. Stuber has continued to perfect the research techniques that are now allowing him and other scientists to home in on the specific cells and cell types that contribute to various neuropsychiatric conditions, such as depression and autism.

[Read University Gazette article on 2014 Hettleman Prize](#)

CIDD Investigator and faculty member, Gabriel Dichter, Ph.D., has been chosen to receive the upcoming 2015 Phillip and Ruth Hettleman Prize for Artistic and Scholarly Achievement by Young Faculty for his advances in understanding neural mechanisms of social motivation in autism. Prior CIDD research faculty award winners include Drs. Mark Zylka and Ayse Belger.

Leah Townsend Receives Weatherstone Autism Speaks Award

Autism Speaks has announced its seventh annual class of Weatherstone Predoctoral Fellows. Fifth year Neurobiology Curriculum student, Leah Townsend, has been selected to receive one of eight Weatherstone Fellowships. Selected from a highly competitive pool of applicants, this predoctoral fellowship program launches promising young scientists into autism careers, pairing them with leading investigators to pursue pioneering research.

Ms. Townsend will investigate how autism-linked gene changes affect the functional development of the brain's cerebral cortex, especially for visual processing. Her goals include the development of detailed profiles of altered brain function that could provide treatment targets for new therapies. Ms. Townsend will work under the mentorship of UNC neuroscientist and CIDD Investigator, Spencer Smith.

Ms. Townsend was also recently recognized at the Triangle Society for Neuroscience (SfN) Spring Neuroscience Meeting where she received the Blackrock Microsystems Travel Award for her poster presentation. The work she presented examined the functional development of cerebral cortex in a mouse model of Angelman Syndrome, implicating the mutation responsible for the syndrome in the development and function of higher order cortical circuits.

Read more about the [Weatherstone Fellowship Program](#).



Improving Education in Co-Occurring Developmental Disabilities and Behavioral Health

CIDD faculty members, Sherry Mergner, MSW, LCSW; Deb Zuver, MA, LMFT; and Morgan Parlier, MSW, LCSW are partnering with the UNC School of Social Work Family Support Program and UNC-PrimeCare to develop specialized education and training for providers who work with individuals who have both developmental disabilities (DD) and behavioral health (BH) issues, and their families. The inaugural North Carolina Area Health Education Centers Program (AHEC) Innovation Fund has been awarded to the Family Support Program to implement this initiative. The project aims to develop a specialized MSW program to prepare social workers to deliver integrated behavioral health services in primary care settings as member of interprofessional teams. The initiative will expand the behavioral health workforce in North Carolina to address the critical health and behavioral health needs of unserved and underserved transitional age youth.

"This is a population whose needs are often overlooked," said Tamara Norris, Clinical Associate Professor and director of the Family Support Program. "At least one-third of youth and young adults who have a developmental disability have co-occurring behavioral health diagnoses, and practitioners haven't been adequately trained to identify the condition and to provide effective interventions and support. This award enables us to add innovative elements to the UNC-Prime Care curriculum, including using the Standardized Patient model approach for skills practice for MSW students in the program, and to have students and service providers learn directly from individuals with disabilities and their families to develop skills in family- and person-centered practice."

CIDD will collaborate with Family Support on the development and production of a DVD of a case study highlighting critical issues related to working effectively with families and adolescents who have DD/BH diagnoses, including effective family/provider partnerships, IP teamwork, and patient- and family-centered practice. The case study will be based on actual experiences of patients in North Carolina's health care system. Actors, family members and individuals with DD/BH will be recruited as Standardized Patients that will offer students with opportunities to develop, practice, and enhance their skills.



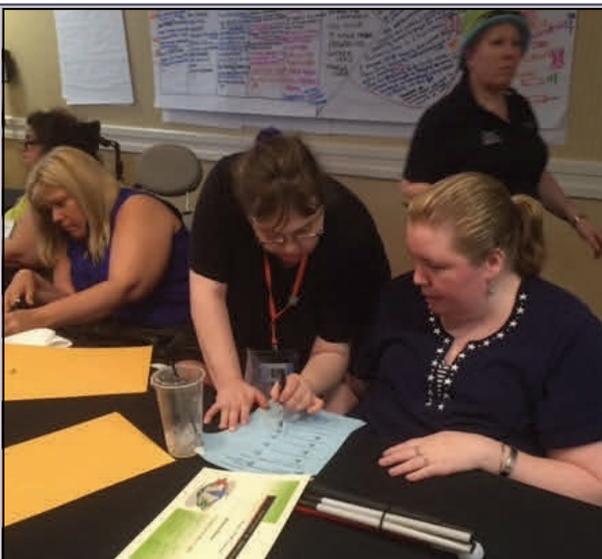
Pictured left to right: Anne Jones, Deb Zuver, Barbara Leach, Tamara Norris, Sherry Mergner, Meryl Kanfer, Morgan Parlier

Maryland Interactive Training in Leadership and Self-Advocacy

The Project STIR (Steps Toward Independence and Responsibility) team comprises four trainers with and without developmental disabilities who present hands-on self-advocacy and leadership training. This spring, the team was invited to Baltimore by the Maryland Developmental Disabilities Administration to train 25 self-advocate leaders recruited from four regions across Maryland.

Project STIR aims for participants to learn to advocate for themselves and mobilize others; they gain leadership experience through four consecutive days of structured interactive workshops and team-building activities. Modules included practice activities with self-awareness, communication, problem-solving steps; and rights and responsibilities. On the final day, participants created a PATH, an accessible strategic planning process, for their work throughout the year ahead.

Project STIR is directed by CIDD's Deborah Zuver; the team includes Robyn Dorton and Wilson Finks. Barbara Leach, Family Support Specialist and trainer at the UNC School of Social Work, presented with the team in Maryland.



Robyn Dorton (pictured center) provides technical assistance to implement the Project STIR approach to building self-advocacy and leadership skills.

Our Social Skills Group for Teenagers and Young Adults With Optional Compensated Research Component Begins Soon!

We are pleased to announce the next session of our group-based social skills clinic for teenagers and young adults. This clinical service consists of 8 hour-long weekly sessions, with the next session scheduled to begin on Monday, September 28th at 4p.m. This clinic is offered through the Carolina Institute for Developmental Disabilities (101 Renee Lynne Court, Carrboro, NC 27510). All major insurance providers are accepted.

Sessions will be led by a clinical psychologist and trainee and will focus on improving social cognition and social skills. The class is open to those with a formal diagnosis that impacts their social abilities (such as autism spectrum disorder) as well as to those simply wishing to improve their social skills. Because much of the class relies on back-and-forth between class members, participants must have sufficient verbal skills to participate in a group setting. Sessions will have up to 12 participants.



Optional Research Component: We will also have an optional research component available for those enrolled in this social skills group that involves three additional visits to the Carolina Institute for Developmental Disabilities. One of these visits will be within two weeks before the first social skills group session, one will be within two weeks after the last social skills group session, and the final visit will be approximately 2 months after the last social skills group session. These research visits will involve a diagnostic interview, questionnaires, neuropsychological testing, and a brief eye tracking session. This first visit will take approximately three hours, for which participants will be compensated \$30; the next two visits will take approximately two hours, for which participants will be compensated \$20 each visit.

All potential patients or caregivers please contact Twyla Peoples at 919-843-1529 or Twyla.peoples@cidd.unc.edu for more information about scheduling and registration.

2-Day Introductory Workshop: Pragmatic Organization Dynamic Display (PODD)

Presented by **Dr. Debbie Reinhartsen**



Pragmatic Organization Dynamic Display

*Multi-Modal Communication Strategies for
Children who have Complex Communication Needs*

Oct 22 and 23, 2015

**Extraordinary Ventures
Chapel Hill, NC**

13 Contact Hours

Children who have complex communication needs or have only limited speech, in addition to other challenges, often struggle to interact and communicate. This may include children who have physical disabilities, multiple disabilities, sensory processing challenges, limited social interaction skills, and/or a range of cognitive limitations and learning difficulties.

This 2-day course will demonstrate the use of a Pragmatic Organization Dynamic Display (PODD) approach developed by Gayle Porter. Generic templates for multi-page "lite tech" communication books have been carefully designed to support genuine communication for a variety of functions throughout the day. These templates may be customized for a range of access methods and other individual needs.

Learn strategies for creating multi-modal language learning environments that provide receptive models and expressive opportunities for language development, as well as, strategies for teaching and using PODD with children and their communication partners. Videos and case examples will be shared. Participants will have an opportunity for hands-on practice with PODD Communication books.

For more information contact Debbie Reinhartsen at (919) 966-4138 or Debbie.Reinhartsen@cidd.unc.edu.

To register online visit: <http://tinyurl.com/pn3wazz>

The Useful Speech Study

Predicting Useful Speech in Children With Autism

A child with “useful speech” can use numerous words meaningfully in different situations to communicate with other people about his needs, wants, and interests. Children with autism spectrum disorder (ASD) who reach or exceed this level of speech development as preschoolers generally have better outcomes in later childhood and as adults than children who do not develop useful speech as preschoolers. So, both because of the immediate advantages of learning to talk and also the importance of useful speech for the future, parents and professionals alike want children with ASD to develop useful speech at as young an age as possible.



Delays in developing useful speech are common in children with ASD. When a child is not talking to communicate by the expected age, two questions often arise for parents: (1) Will my child learn to talk? (2) What can I do to help my child learn to talk? The Useful Speech Study tried to shed some light on such questions by studying young children with ASD who started the study when they did not yet have useful speech, and following each child for 16 months to learn about the way they developed.

Earlier research had identified a number of behaviors or skills in young children with ASD or behaviors of parents and other caregivers that might help children develop useful speech. The Useful Speech Study looked at nine of these skills or behaviors: (1) looking at other people who are speaking in a child-friendly way; (2) looking at things that other people show or point to; (3) understanding spoken words; (4) showing intent to communicate with other people by using gestures, gaze, and vocalizations; (5) using different consonant sounds in communicative vocalizations (even non-words); (6) imitating actions of others; (7) oral-motor skills, as seen in the child’s eating and drinking; (8) playing with objects appropriately in varied ways; and (9) parents using language to respond to their children’s play and communication. We were interested in finding which of these 9 variables were the best predictors of the children’s development of useful speech.

We found that four variables together gave us the most information about the children’s development of useful speech: (a) looking at things that other people show them or point to; (b) showing intent to communicate with other people by using gestures, gaze, and vocalizations; (c) using different consonant sounds in communicative vocalizations, even before the child is using any words; and (d) parents using language to respond to their children’s play and communication. Thus, although we cannot definitively answer the question, “Will my child learn to talk?” we can say that when these four things happen frequently before a child has developed useful speech, he will be more likely to develop useful speech over the next year or so. Similarly, the Useful Speech Study does not give definite answers to “What can I do to help my child learn to talk?” But focusing on these four variables (3 child behaviors and 1 adult behavior) in interventions for children with ASD who have not yet developed useful speech may be especially helpful. Hopefully, future research will test these ideas in intervention studies.

The Useful Speech Study was a joint project between Vanderbilt University and UNC-CH, with Dr. Paul Yoder at Vanderbilt serving as the Principal Investigator for the whole study, and Dr. Linda Watson serving as the site Principal Investigator at UNC-CH. This research was funded by National Institute for Deafness and other Communication Disorders (NIDCD R01 DC006893) and supported by the National Institute for Child Health and Disorders (NICHD) through the Vanderbilt Kennedy Center (P30HD15052) and the Carolina Institute for Developmental Disabilities (P30HD03110), which assisted us in recruiting the NC participants through the Autism Research Registry. We are very grateful for our wonderful staff, and especially for the families who trusted us with their precious children.



Global Summit on Innovations in Health and Intellectual and Developmental Disabilities

Pamela Smith, MSN, 2014 graduate of the North Carolina Leadership Education in Neurodevelopmental Disabilities (LEND) program and Michelle Franklin, MSN, NC LEND community nurse practitioner faculty member, attended the American Academy of Developmental Medicine and Dentistry (AADMD) Global Summit on Innovations in Health and IDD Conference in Los Angeles, CA July 27-29, 2015.



On July 27th, Ms. Smith and Ms. Franklin participated in the poster presentation communicating the work in which they, along with Rob Christian, MD, Associate LEND Director, have been engaged, titled "Developing an Intellectual and Developmental Disabilities Specific Curriculum for North Carolina Nurse Practitioners." Their poster presentation highlighted CIDD's commitment in equipping nurse practitioners with advanced leadership skills and specialized training to provide evidence-based developmental disabilities care. Ms. Smith, Ms. Franklin, and Dr. Christian were awarded 2nd place for this poster presentation.

Pictured from right to left: Dr. Matthew Holder (President of AADMD), Michelle Franklin, Pamela Smith, Justin Gentry, Dr. Nancy Dougherty

Krysta Gougler-Reeves Attends Disability and Policy Seminar in Washington, DC

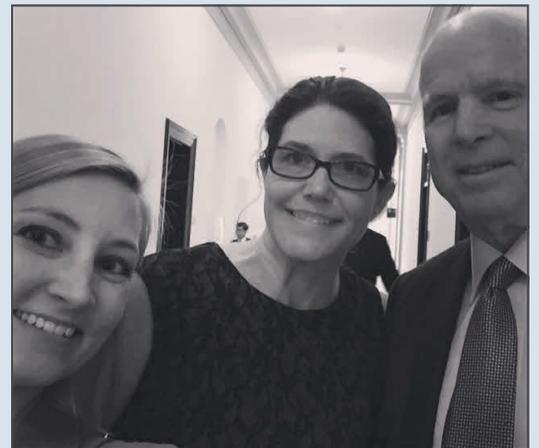
Krysta Gougler-Reeves is a dual degree candidate at UNC School of Social Work and UNC Gillings School of Global Public Health. During the 2014-15 academic year Ms. Gougler-Reeves completed her social work field placement at the CIDD and was also involved in the Leadership in Neurodevelopmental and Related Disorders (LEND) training program.

This past April, Ms. Gougler-Reeves represented our LEND program at the Disability Policy Seminar held in Washington, D.C. The conference centered around learning about critical programs such as SSI, SSDI, Medicaid, and home based supports that support people with disabilities. Professionals, students, and self-advocates were in attendance.

The last day of the conference was reserved for attendees to make visits to Capitol Hill to speak with their representatives. "I teamed up with several self-advocates from the Arc and met with staffers of Senator Burr (who was instrumental in helping with the ABLE Act), and Rep. Butterfield," shared Ms. Gougler-Reeves. "It was powerful to speak with our representatives alongside self-advocates who shared their stories."

"While we were there, we met Senator John McCain in the hallway and took the opportunity to shake his hand, introduce ourselves, and explain that we were a group from North Carolina with the Disability Policy Seminar who planned to talk with our representatives and advocate for the rights of people with disabilities," added Ms. Gougler-Reeves. "Naturally, I had to document the encounter with a selfie."

This academic year, Ms. Gougler-Reeves will serve as graduate student coordinator for UNC's Maternal and Child Health (MCH) Leadership Consortium. CIDD's LEND program has been involved in this leadership training program since its inception. This integrated approach to interdisciplinary training has become a national model and serves as an effective platform for academic – practice engagement in public health practice in MCH.



Pictured left to right: Krysta Gougler-Reeves and fellow conference attendee, Kelley Stadler, with Senator John McCain

2015 CIDD Community Talk Series

The CIDD hosts a series of talks to share information about recent advances in developmental disabilities. These sessions are a great opportunity for parents, teachers, professionals, and others to learn more about specific developmental disabilities topics. All talks are free, and everyone is welcome.

Wednesday, September 9th

Ann Turnbull, Ed.D.

Distinguished Professor Emerita, Department of Special Education,
University of Kansas; Adjunct Professor, School of Education, University of North Carolina

**The “Nuts and Bolts” for a Person with Significant and Multiple Disabilities
to Have a Home of His Own: The Turnbull Family’s Experience**

Dr. Turnbull will share how she and her husband, Rud, created a home for their son, Jay—finances; recruiting, hiring, and managing housemates; location, floor plan; daily activities, and more.



Wednesday, October 14th

William J. Hussey, M.A.

Director, Exceptional Children Division, DPI

**What Families and Educators Need to Know about Common Core
to Support a Child with an IEP**

This presentation will discuss how standards, common core or state standards, need to be broken down to address their underlying skills. It is the specific skills needed to address any standards that are used to develop a child’s IEP.



Wednesday, November 11th

Anthony D. Nicholson, Esq.

Attorney

**The Role of Special Needs Trusts in Estate Planning for Families
with Children with Developmental Disabilities**

Mr. Nicholson will outline the types of estate planning documents that are important for families with children who have developmental disabilities, with an emphasis on special needs trusts.



Wednesday, December 9th

Tracy Vail, M.S., CCC/SLP

Speech/Language Pathologist, Autism Consultant

Functional Communication Training

Ms. Vail will discuss how to determine the meaning of a child’s maladaptive behavior and then how to teach the child to use more effective communication forms to meet their needs.



Sessions are held from 6:30PM to 8:00PM at the CIDD Conference Room 101.

To RSVP or for more information, please contact:

Debbie B. Reinhartsen at (919) 966-4138 or Debbie.Reinhartsen@cidd.unc.edu

Appreciation for the CIDD Information Technology Team



UNC
CAROLINA INSTITUTE FOR
DEVELOPMENTAL DISABILITIES



*CIDD Information Technology team pictured left to right:
Brian Wrighten, Ernest Clemons, Tom Gray*

The CIDD Information Technology (IT) team plays an integral role in how the CIDD functions across its core areas of services, training/education, research and dissemination. The IT staff is comprised of Tom Gray, Brian Wrighten and Ernest Clemons. All are highly trained and have achieved industry recognized certifications including ones from Cisco, Microsoft and Dell. The IT team have a combined 50+ years of experience in the IT field and have received numerous awards for service excellence.

Managing the IT needs of the CIDD is no small task. Our IT team supports more than 200 users working on over 150 computers; 12 servers with over 30 Terabytes of secure disk storage, and several large databases; and also developed and maintain over 15 websites.

Our IT team has led the campus in several areas of innovation. They have established a secure remote access solution for employees to access work files from remote locations; instituted a remote support capability to diagnose and repair desktop issues, and established a digital clinic where all of our clinic rooms have been outfitted with cameras and microphones to enhance the teaching and training missions of the CIDD.

Finally, and perhaps most importantly, the CIDD IT team is readily available to assist faculty, staff and trainees with day-to-day questions and IT needs. The IT response is timely, effective and delivered with patience and professionalism. The CIDD is very fortunate to have such a talented IT team!

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!

A gift to the Carolina Institute for Developmental Disabilities is an investment in the lives of thousands and in the future of our communities. Join us by giving today. To make a donation by credit card, please visit the Medical Foundation of North Carolina's gifting page and choose "Carolina Institute for Developmental Disabilities:" [Click Here.](#)

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Newsletter Editor—Keath Low