Baby Teeth and Autism: UNC Part of Large NIH Study of Environmental Risk Factors

As part of the new $157 million NIH ECHO initiative, a research network led by UNC’s Joseph Piven, MD, director of the Carolina Institute for Developmental Disabilities, will join researchers across the country in a study to explore what baby teeth can tell us about a child’s risk of developing autism if the child was exposed to chemicals in the womb.

Drexel University’s Craig Newschaffer, PhD, is the principal investigator. To conduct the study, his team will work with the NIH Autism Center of Excellence directed by Piven, the Thomas E. Castelloe Distinguished Professor of Psychiatry in the UNC School of Medicine.

“This study will allow us to examine the impact of exposure to environmental toxins at various times during a mother’s pregnancy on the gradual unfolding of autism in the first few years of life,” Piven said. “UNC has led a national network of investigators, as part of the Infant Brain Imaging Study (IBIS), in examining early behavior and early brain features that unfold as infants begin to show evidence of autism at age 2 or 3. We are a leader in this field and proud to be part of the NIH ECHO Initiative.”

The study will focus on infants who are at high risk for autism by virtue of having an older sibling with autism, as those infants have a substantially higher risk of developing autism by the time they are 2 to 3 years old. Newschaffer, founding director of the A.J. Drexel Autism Institute, hopes to use the shed baby teeth to examine whether environmental factors play a role in further increasing that risk.

This team will study a group of 1,713 subjects called the Autism Spectrum Disorder Enriched Risk (ASD-ER) cohort. In this cohort, there are 1,281 children who are considered “high risk” because of their older sibling’s autism diagnosis, and 432 children called “low risk” because they lack a sibling with autism.

Newschaffer and his co-investigators will test lost baby teeth for levels of environmental chemicals that the children would have been exposed to while in their mother’s womb. That time is particularly important for neurodevelopment and, thus, particularly vulnerable.

Chemicals being examined in the study include organochlorine pesticides, such as DDT, and polychlorinated biphenyls, formerly used for things like insulating electrical transformers. Although banned in the 1970s, the chemicals, true to their “persistent” moniker, remain in the environment and human exposure continues. The study will also investigate phthalates, which were used to make plastics more flexible.

The researchers hope to determine whether prenatal exposure to these chemicals increases the risk of autism. Additionally, for a smaller group of participants where genetic data are already available, the researchers hope to explore whether the genetic susceptibility of a child plays any role on the effect chemical exposures have on autism risk. Findings will be useful in determining the mechanisms underlying autism. Once such mechanisms are better understood, more effective prevention and treatment strategies can be developed.
Please join us in congratulating Dr. Greg Olley on his retirement. Dr. Olley has spent almost 30 years at UNC and has made significant contributions to the field of intellectual and developmental disabilities. At the Center for Development and Learning/Carolina Institute for Developmental Disabilities, Dr. Olley has served as interim Director, UCEDD Director, clinical psychologist, teacher and supervisor/mentor to many graduate students and community providers. From a statewide perspective, he has served on many committees and boards such as the Governor’s Commission for Mental Health, Developmental Disabilities and Substance Abuse Services, the NC Developmental Disabilities Council and has partnered closely with Disability Rights NC for many years. Renewal of our LEND grant will enable us to continue our work for another five years along with our partners at UNC-Greensboro, Duke, North Carolina Central University, North Carolina State University, as well as state agencies including the Department of Health and Human Services and the Department of Public Instruction.” LEND is one of several initiatives funded by federal Autism CARES legislation.

Congratulations to Dr. Greg Olley on His Retirement

Dr. Greg Olley

Baby Teeth and Autism: UNC Part of Large NIH Study of Environmental Risk Factors continued

“We’re very excited about the prospect of applying this novel approach to measuring prenatal environmental exposure in larger groups of children in ECHO,” Newschaffer said. “We’d love to integrate this with available genomics measures to, at a scale that has yet to be approached, study the interaction between genetics and environment.”

The NIH awarded $157 million for many projects under ECHO, which will investigate how exposure to environmental factors in early development — from conception through early childhood — influence the health of children and adolescents. “Every baby should have the best opportunity to remain healthy and thrive throughout childhood,” said Francis S. Collins, MD, PhD, NIH director. “ECHO will help us better understand the factors that contribute to optimal health in children.”

Piven’s Infant Brain Imaging Study includes collaborators at UNC, Washington University, the University of Washington, and the University of Pennsylvania/Children’s Hospital of Philadelphia.

More information is available at the NIH Echo initiative website and in the NIH press release.
Miami Professor Bicycles to Chapel Hill to Raise Awareness for Angelman Syndrome

In August, after cycling 1,000 miles from Miami to Chapel Hill, Kellen Hassell, EdD, presented a check for $15,000 to the Carolina Institute for Developmental Disabilities (CIDD) earmarked for research on Angelman syndrome, a severe neurodevelopmental condition that occurs in about one in 15,000 live births. He chose Chapel Hill because of UNC’s dedication to patients with Angelman syndrome and research aimed at creating better treatments.

“This is where there is not only one of the very first interdisciplinary health clinics designed to diagnose and treat Angelman syndrome and support parents and families who have a loved-one with the condition,” Hassell said on his fundraising page, “but also where some of the most promising cure research is being conducted by Dr. Ben Philpot and his team.” The total sum raised thus far exceeds $18,575.

Hassell is clinical assistant professor at the Nicole Wertheim College of Nursing and Health Services at Florida International University in Miami. He and his wife have three young children. The middle child, Luc, who is four years old, has Angelman syndrome. He has severe intellectual disabilities, trouble sleeping, and other developmental problems. He may never understand what his father did to raise awareness and money for research.

Hassell, no stranger to raising money for causes, handed an oversized check to UNC researchers at a ceremony following his journey. Mark Zylka, PhD, member of the CIDD and director of the UNC Neuroscience Center, Philpot who serves as associate director, and Joseph Piven, MD, director of the CIDD, expressed their gratitude for this generous contribution.

“Dr. Hassell’s cycling trek was truly amazing and inspiring, and it helps to keep us all focused on the goal of developing new treatments for children and adults with Angelman syndrome,” Zylka said.

To donate, click here.

To read more about Hassell’s efforts and to watch him speak about his son and the potential for research breakthroughs, click here or read the News & Observer story here. Read more about Philpot’s latest work here.

Researchers in the labs of Ben Philpot, PhD, Spencer Smith, PhD, and Mark Zylka, PhD, thank Dr. Kellen Hassell and the Angelman Syndrome Foundation for their support.
Tricks to Treat Angelman Syndrome May Lie in ‘Clock Cells’

A newly discovered phenomenon in cells that regulate the sleep-wake cycle may provide clues for how to treat Angelman syndrome and dup15q syndrome, two autism-related conditions. The report was published 16 June in *Scientific Reports*.

Both syndromes involve alterations to UBE3A. Researchers had thought that this gene is expressed from only the copy of chromosome 15 inherited from the mother, and that the paternal copy of UBE3A is silent, via a phenomenon known as imprinting. The new study shows that some neurons in the brain do express the paternal copy of the gene. These neurons are in the suprachiasmatic nucleus, a ball of cells buried deep in the brain that serves as the time-keeper for the body’s circadian, or sleep-wake, cycle.

In Angelman syndrome, the maternal copy of UBE3A is mutated, leading to developmental delays, epilepsy and problems with speech. Dup15q syndrome, by contrast, stems from a duplication of a segment of the maternal chromosome that includes UBE3A. Children with dup15q may have autism, as well as severe seizures and cognitive problems.

Researchers are working on treating Angelman syndrome by reanimating the paternal copy of UBE3A. Understanding how certain neurons still express the paternal copy of the gene could help researchers develop treatments that have few side effects, says lead researcher Benjamin Philpot, professor of neuroscience at the University of North Carolina at Chapel Hill.

“If we understand more about the mechanism of imprinting, we might get some greater insights into how to specifically regulate it,” he says. “The mechanism might also point to a way of silencing the extra maternal copy in dup15q syndrome.”

Youthful Cells:

The findings underscore the complexity of how the brain controls UBE3A, says Stormy Chamberlain, assistant professor of genetics and genome sciences at the University of Connecticut in Storrs, who was not involved in the work. “In the past, we painted the story in black and white: Neurons imprinted UBE3A and non-neurons didn’t,” she says. “This study takes the story up a level by showing that only certain types of neurons are imprinted.”

Last year, Philpot and his team reported hints that neurons in the suprachiasmatic nucleus express paternal UBE3A. The new work confirms this idea. The neurons that express paternal UBE3A tend to have markers of immaturity. This observation is consistent with the fact that imprinting ramps up after birth, and that immature neurons in newborn mice express paternal UBE3A. In people, neurons mature later than they do in rodents, so even more neurons may express the paternal copy than in mice, says Chamberlain. “I’m curious how well this finding extends to human brains,” she says.

On the Clock:

Many of the subset of neurons in the suprachiasmatic nucleus that express UBE3A are also circadian clock cells. The researchers found that neurons in other brain regions involved in circadian rhythms also express low levels of paternal UBE3A.

Children with Angelman syndrome often have a lot of trouble falling asleep, which is in part regulated by the circadian clock. But the presence of UBE3A in circadian clock cells suggests that these sleep challenges arise elsewhere.

The study hints that there is a relationship between immaturity, the circadian clock and the lack of imprinting, says Janine LaSalle, professor of medical microbiology and immunology at the University of California, Davis, who was not involved in the work. The next step, Philpot says, is to identify the genes that shut down UBE3A by comparing neurons that have the silenced gene with those that escape imprinting. That finding could allow researchers to devise treatments that involve manipulating the imprinting process.

*This article written by Jessica Wright was originally published in Spectrum on July 4, 2016. It has been reprinted with permission. View original at this link: https://spectrumnews.org/news/tricks-to-treat-angelman-syndrome-may-remote-in-clock-cells/*
In August of 2016 the Pitt Hopkins Research Foundation awarded its first gene therapy grant, to Drs. Steve Gray and Ben Philpot at the University of North Carolina at Chapel Hill. The laboratories of Dr. Philpot and Dr. Gray are collaborating on a project to investigate the feasibility of a gene therapy approach for Pitt-Hopkins syndrome (PTHS). This collaborative study combines Dr. Philpot's expertise in autism and neuroscience with Dr. Gray's expertise in translational gene therapy for neurological disorders. The project will follow a platform gene transfer approach using AAV vectors taken by Dr. Gray to initiate a human Phase I trial for Giant Axonal Neuropathy. The approach uses an engineered virus, AAV, to carry a functional copy of the gene disrupted in PTHS into the body and distribute it across the nervous system. In this fashion, a single dose of this gene therapy could permanently restore the gene to cells across the nervous system, treating the disease at its source. This initial pilot study is meant to assess the potential of this as a treatment approach for PTHS, and identify any roadblocks that may exist.

Dr. Mark Zylka Receives Angelman Syndrome Foundation Award

The Angelman Syndrome Foundation has funded three groundbreaking research proposals, each focusing in a different and very important area of AS research. One of these studies led by CIDD Investigator, Dr. Mark Zylka, seeks a further understanding of the complexities of Ube3a and how the paternal copy can best be reactivated.

Based on previous research conducted at Dr. Zylka's lab and Dr. Ben Philpot's lab, Dr. Zylka's team will continue this work with the goal of unsilencing the paternal copy of Ube3a using new methods. Past research has shown that topoisomerase inhibitors, a drug compound, can unsilence the paternal copy of Ube3a, but that this drug also affects a number of other genes. Dr. Zylka's team will use new technology (CRISPR/Cas9 screening technology) and a new approach to unsilence Ube3a, called a genetic screen. We know there are more than 20,000 genes in the human genome, and this genetic screen will evaluate each of those genes to see if any of them can unsilence the paternal copy of Ube3a. This is a novel new approach that has not been conducted before. In the end, the research team hopes to have a list of genes that can unsilence paternal Ube3a with little to no impact on other genes during the process.

This research is important because we know from previous research that unsilencing Ube3a using specific drugs can affect a number of other genes in the process. This research, using CRISPR/Cas9 screening technology and a new approach to unsilencing Ube3a, will look for genes—rather than drugs—that can unsilence paternal Ube3a with little to no impact on other genes during the process.

Click here to watch video and hear directly from Dr. Zylka about the research project.

Drs. Steve Gray and Ben Philpot Receive Pitt Hopkins Research Foundation Award

In August of 2016 the Pitt Hopkins Research Foundation awarded its first gene therapy grant, to Drs. Steve Gray and Ben Philpot at the University of North Carolina at Chapel Hill.

The laboratories of Dr. Philpot and Dr. Gray are collaborating on a project to investigate the feasibility of a gene therapy approach for Pitt-Hopkins syndrome (PTHS). This collaborative study combines Dr. Philpot's expertise in autism and neuroscience with Dr. Gray's expertise in translational gene therapy for neurological disorders. The project will follow a platform gene transfer approach using AAV vectors taken by Dr. Gray to initiate a human Phase I trial for Giant Axonal Neuropathy. The approach uses an engineered virus, AAV, to carry a functional copy of the gene disrupted in PTHS into the body and distribute it across the nervous system. In this fashion, a single dose of this gene therapy could permanently restore the gene to cells across the nervous system, treating the disease at its source. This initial pilot study is meant to assess the potential of this as a treatment approach for PTHS, and identify any roadblocks that may exist.
Carolina Institute for Developmental Disabilities (CIDD) Investigator Dr. Gabriel Dichter was awarded a grant from the National Institute of Mental Health to evaluate dopaminergic functioning during reward processing in individuals with autism spectrum disorder. This study represents the first ever NIH-funded simultaneous PET/MR study at UNC and the first PET study of dopamine signaling in autism using the radiotracer raclopride. The UNC team includes postdoctoral fellow Dr. Jessica Kinard, BRIC Director Dr. Weili Lin, Cyclotron and Radiochemistry Research Program Director Dr. Zibo Li, CIDD faculty Dr. Heather Cody Hazlett, and CIDD Director Dr. Joe Piven.

This grant will use a new brain imaging scanner at the Biomedical Research Imaging Center (BRIC), a Siemens Biograph mMR, which represents a major technological breakthrough in the imaging field by combining positron emission tomography (PET) and magnetic resonance imaging (MRI) in one scanner. Through the acquisition of simultaneous PET and fMRI while research participants perform a reward task, this study will evaluate relations between dopamine signaling (derived via PET) and brain activation (derived via fMRI) during reward processing in autism. The simultaneous acquisition of PET and fMRI combines the detailed anatomical information offered by an anatomical MRI scan, the high sensitivity to in vivo molecular events from a PET scan, and the high temporal resolution of a functional MRI scan.

The other PI of this project is Dr. Jacob Hooker, the Director of Radiochemistry at the Martinos Center for Biomedical Imaging and Associate Director of the PET Core at Massachusetts General Hospital. Dr. Hooker is an expert in clinical PET neuroimaging and was the guest speaker at the 2015 annual CIDD IDDRC retreat. His lab will provide expertise in PET data acquisition and analysis to compliment Dr. Dichter’s expertise in functional brain imaging in autism.

This grant aims to understand the contribution of impaired dopamine signaling to reward processing impairments in autism. There is a large preclinical research literature that implicates impaired dopamine functioning in the etiology of autism, and additionally there is fMRI evidence indicative of compromised brain activation in response to social rewards in autism. However, since the source of fMRI signals is nonspecific, the contribution of impaired dopamine signaling to these fMRI patterns in autism is unknown. This study represents a critical first step to understanding linkages between dopamine signaling during reward processing, brain hypoactivation during reward processing, and symptom expression in autism.

This project will also set the stage for future translational autism studies at UNC. There is an urgent need to speed the discovery of new autism treatments through pipelines that evaluate the potential for novel compounds to rescue the functioning of relevant brain targets in autism. Whereas preclinical research is capable of delineating molecular mechanisms that are impaired in animal models of autism, traditional fMRI has been limited in its capacity to leverage this information to evaluate whether similar molecular systems are impaired in autism, and, most critically, whether novel compounds are capable of rescuing such systems in autism. Due to the molecular specifically of PET afforded through the use of specific ligands, PET has the capacity elucidate novel neural targets for future experimental therapeutic autism clinical trials. This project will help to establish PET expertise at UNC to allow future translational studies that combine preclinical and clinical PET studies to evaluate promising new autism treatments.
Researchers at the University of North Carolina (UNC) and the University of Minnesota (UMN) have been awarded a $4 million grant from the National Institutes of Health (NIH) to launch the Baby Connectome Project (BCP). The BCP aims to provide scientists with unprecedented information about how the human brain develops from birth through early childhood and will uncover factors contributing to healthy brain development.

“This is an unprecedented effort to map the development of brain circuits during a stage when our brains undergo highly dynamic changes that have life-long impacts on cognitive development. We are thrilled to have the opportunity to carry out this exciting project,” said Weili Lin, PhD, Dixie Soo Distinguished Professor in Neurological Medicine at the UNC School of Medicine, director of the UNC Biomedical Research Imaging Center (BRIC), professor and vice chair of Basic Research, Radiology and contact principal investigator of the Baby Connectome project.

The BCP is a four-year research initiative of the NIH, supported by Wyeth Nutrition, through a donation to the Foundation for the National Institutes of Health (FNIH). UNC and UMN are the recipients of the NIH grant. The collaboration will bring together the Biomedical Research Imaging Center (BRIC) at UNC and the Institute of Child Development (ICD) and Center for Magnetic Resonance Research (CMRR) at UMN. Both institutions have substantial expertise in infant/pediatric neuro-imaging and behavioral assessment.

The project will characterize human brain connectivity and map patterns of structural and functional connectivity to important behavioral skills from infancy to early childhood. Additional biological (e.g., genetic markers) and environmental measures (e.g., family demographics) will be collected and examined to provide a more comprehensive picture of the factors that affect brain development. Findings from this study will provide other scientists with a definitive foundation to inform new questions about typical and atypical brain and behavioral development. Additionally, this study promises to inform policy decisions that could directly or indirectly affect healthy brain development during early childhood.

“The UNC/UMN team is uniquely suited to perform this challenging but critical task, and we expect the data collected and results that come from the BCP to have broad implications for understanding the most dynamic period of human brain development,” said Jed Elison, PhD, a co-principal investigator of the BCP from UMN, and former post-doctoral student at UNC.

“Wyeth Nutrition is excited to support research at UNC and UMN through our partnership with the FNIH,” said CEO Wyeth Nutrition Mike Russomano. “This innovative research – led by two institutions at the forefront of studying brain development in children – will add to a better understanding of what is needed to support brain development, and the overall health of infants and children, in the critical first years of life.”

For the project, researchers at UNC and UMN will perform safe and non-invasive magnetic resonance imaging (MRI) brain scans of 500 typically developing children, ages 0-5 years, over the course of four years. Of these 500 children, 175 will come from two other brain imaging studies already underway, one from UNC (“Early Brain Development in 1 and 2 Year Olds,” led by John Gilmore, MD) and one from UMN (“Infant Brain and Behavioral Signatures of Later Emerging Risk for Psychopathology,” led by Jed Elison, PhD). All of the data collected will be shared with the broader scientific community to accelerate discovery.

The project will include longitudinal groups, where children will be scanned four to six times at different ages, and cross-sectional groups, where they will be scanned once at distinct points in their development. In addition to the imaging data collected, researchers also will obtain parent reports and direct assessment of cognitive and behavioral development in the participating children. All of the collected information will inform a more comprehensive picture of how emerging patterns of brain connectivity shape behavioral development in children under the age of 5.

UNC and UMN will leverage technological innovations developed through the original Human Connectome Project (HCP), a scientific endeavor funded by the NIH to create a map of the circuitry within the human brain, to investigate the structural and functional changes that occur during typical development. This project will be part of the Lifespan Human Connectome Project (L-HCP), which aims to extend the HCP to map connectivity in the developing, adult, and aging human brain. It is funded by the NIH Blueprint for Neuroscience Research, a collaborative framework through which 15 NIH institutes, centers and offices jointly support neuroscience-related research, with the aim of accelerating discoveries and reducing the burden of nervous system disorders.

CIDD Investigators participating in the project include John Gilmore, MD (co-PI); Heather Cody Hazlett, PhD; Joe Piven, MD (co-PI); and Martin Styner, PhD.
Dr. Laura Politte Receives North Carolina Translational and Clinical Sciences Institute Award

Laura Politte, MD, has been awarded a position in the North Carolina Translational and Clinical Sciences (NC TraCS) Institute KL2 Program. The KL2 is a career development award that provides salary support, training in research methodology, formal mentorship, and funding for clinical research for up to 3 years. The program is funded by the National Institutes of Health and administered by the UNC Clinical and Translational Science Award center. The grant will allow Dr. Politte to continue patient care 25% of the time and devote 75% time to research efforts. As part of this research award, Dr. Politte will be conducting a randomized, placebo-controlled trial of sulforaphane, a compound derived from broccoli extracts, that has shown promise as a safe and well-tolerated intervention for improving symptoms of autism in another study. Sulforaphane increases antioxidant levels and can reduce inappropriate inflammation in the body, which has been observed in some individuals with autism. Sulforaphane has also been studied for a number of other conditions, including breast cancer, prostate cancer, and the harmful effects of air pollution. This research study is supported by a collaborative grant from UNC’s Nutrition Research Institute in Kannapolis, NC.

Dr. William & Enid Rosen Research Award, presented at the 15th International Fragile X Conference in San Antonio, Texas. The award is given once every two years by the National Fragile X Foundation to a researcher who has made outstanding contributions to our understanding of Fragile X. Dr. Wheeler is a licensed psychologist and research public health analyst at RTI International. She is also an Adjunct Faculty member at the Carolina Institute for Developmental Disabilities (CIDD). Dr. Wheeler has more than 15 years of research and clinical experience focusing on individuals with intellectual or developmental disabilities and their families. She is an expert in the assessment of children with disabilities, has led the clinical assessment teams for numerous research projects, and has visited more than 100 families of children with FXS to conduct child assessments during home visits. Dr. Wheeler has published more than 20 articles on fragile X syndrome, focusing primarily on behavioral and social/developmental and temperament in young children, family and maternal well-being, and mother-child interaction patterns. Her publications have been especially important in providing new information about the experiences and health outcomes of women with the FMR1 premutation and the relationship between autism and fragile X syndrome. She played a major role on the team that established Our Fragile X World, a survey research registry for families of children with FXS. In addition to her work in fragile X syndrome, Dr. Wheeler is an expert in Prader Willi syndrome and coordinates a major clinic for children with Angelman syndrome at the CIDD.

Dr. Meghan Swanson Receives Pathway to Independence Award

Dr. Meghan Swanson has been awarded an NIH Pathway to Independence Award (K99/R00) from the National Institute of Mental Health to explore the potential protective effects of the early language environment on brain and behavior development during infancy, a period of time before many of the diagnostic features of autism have emerged and when the brain is capable of immense plasticity. It is currently unknown if the early language environment from birth to two-years interacts with intrinsic genetic risk susceptibilities to influence brain and behavior development in infants at high familial risk for autism. To begin to answer these questions infants will participate in multiple brain scans, and home language recordings will be acquired using pocket-sized recorders developed by the LENA Research Foundation. Dr. Swanson will then examine if the home language environment impacts brain and behavioral development in infants who go on to have autism. Her work is focused on data collected from the NIH ACE Network, Infant Brain Imaging Study, a longitudinal neuroimaging study of infants at high and low familial risk of autism, which is led by Dr. Piven. The five-year Pathway to Independence Award provides training and funding for two years while Dr. Swanson continues her postdoctoral fellowship working with Drs. Joseph Piven and Martin Styner, followed by an additional 3 years of funding as she launches her independent research career.
Promising Results with New Gene Therapy Approach for Treating Inherited Neurodegenerative Diseases

A new gene therapy approach designed to replace the enzyme that is deficient in patients with Tay-Sachs and Sandhoff diseases, inherited neurodegenerative disorders, successfully delivered the therapeutic gene to the brains of treated mice, restored enzyme function, and extended survival by about 2.5-fold.

The implications of these promising results for developing similar gene therapies for use in humans and for targeting additional brain disorders are reported in two articles published in the journal, Human Gene Therapy.

Steven Gray, PhD from the University of North Carolina School of Medicine, and Jagdeep Walia from Queen's University in Kingston, Canada, led a team of researchers in the successful development of a specialized adeno-associated virus (AAV) vector. The vector was designed to deliver a gene coding for portions of the alpha and beta subunits of the enzyme that are defective in the Tay-Sachs and Sandhoff mice, respectively. The novel gene transfer vector, administered intravenously, was able to deliver the therapeutic gene to the brain and spinal cord, the targeted sites of action.

“I'm proud of the highly collaborative nature of this work that made it possible, between three institutions across the U.S. and Canada, and in partnership with the New Hope Research Foundation,” said Gray, an assistant professor in the Department of Ophthalmology and a researcher in UNC's Gene Therapy Center and Carolina Institute for Developmental Disabilities. “Several cutting-edge technologies came together on this, which can now be applied to multiple other rare nervous system diseases. In the meantime, our goal is to develop a human treatment for both Tay-Sachs and Sandhoff diseases based on this work.”

“This important proof-of-concept study sheds important information on the optimal design of rAAV vectors for this class of disorders,” says Human Gene Therapy Editor-in-Chief Terence R. Flotte, MD, Celia and Isaac Haidak Professor of Medical Education and Dean, Provost, and Executive Deputy Chancellor, University of Massachusetts Medical School, Worcester, MA.

Dr. Steven Gray Recipient of Health Care Heroes Award

The Triangle Business Journal named its 2016 Health Care Heroes. UNC Medical Center had five honorees including CIDD Investigator, Dr. Steven Gray.

Dr. Gray led a team of researchers at the UNC School of Medicine that developed an innovative, experimental gene transfer-based treatment for children with giant axonal neuropathy (GAN). A clinical trial of this treatment is now underway at the National Institutes of Health (NIH) in Bethesda, Maryland. 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.

Dr. Steven Gray (far left), Award Recipient with Dr. Aziz Sancar, Award Recipient and Nobel Laureate
Self-Advocate Leaders: Paul Offen and Kenneth Kelty

The CIDD leads the country in including self-advocates in LEND graduate-level training. Similar models are now offered at other universities. One or two self-advocate trainees participate during each academic year in the course (Developmental Disabilities Across the Lifespan) and complete additional projects required of all LEND trainees. In addition, self-advocates participate in leadership development, complete community outreach, become members in the NC Postsecondary Education Alliance, and present individual projects at the end of the academic year. Deborah Zuver and Donna Yerby mentor the trainees. McCafferty is their education coach.

Paul Offen and Kenneth Kelty were interviewed about their experiences in LEND Leadership training for 2015-16.

Paul Offen holds a BS in Mathematics from NC State and he participated in previous projects at UNC-TEACCH. One of his goals as a LEND trainee was to better understand the perspectives of others and to contribute his own perspective in the interdisciplinary opportunities at CIDD. Mr. Offen stated that the Leadership Consortium was a highlight of his experience, as he learned more about himself and others. The graduate level problem-based learning course provided increased awareness of other populations and he enjoyed the class discussions. Mr. Offen was already an advocate but he believes he has increased his commitment and his skills in the past year. He presented at several conferences and was on a panel at the annual Northwest Autism Conference in Winston-Salem. Mr. Offen’s goals include applying his leadership experiences on the job and seeking additional experiences to advocate for others.

Kenneth Kelty is a graduate of the University Participant program at Western Carolina University and he is employed part-time at the Arc of Wake County. As a LEND trainee, Mr. Kelty participated in Project STIR (Steps Toward Independence and Responsibility), presenting workshops and talks in high schools, universities, and community settings. Mr. Kelty states that he gained more knowledge and improved his critical thinking, research, writing, and public speaking skills through his experiences at the CIDD. He has written and blogged about his “journey with inclusion” and the need to “broaden the lens of advocacy.” He hopes to obtain a related position in Washington. Mr. Kelty is currently working on a book. His story and his blog can be found on the Autism Speaks website.

McCafferty Hired as Advocacy Liaison

The CIDD is pleased to announce the launch of an exciting new LEND (Leadership and Education in Neurodevelopmental Disabilities) position, that of Advocacy Liaison. Several years ago, Family Faculty was added, but until recently the CIDD has lacked a similar role to include a self-advocate perspective. This fall, S. Wright Kermon, who goes by only McCafferty, was recruited to fill this new position. She made outstanding contributions as a LEND trainee, a LEND fellow, and as a participant in the MCH Leadership Consortium. While doing this work, she developed a Coaching Approach perspective for educational support which she will continue to provide to the LEND self-advocate trainees within our inter-professional cohort. As Advocacy Liaison, McCafferty’s role will expand to include participation in advisory committees, technical assistance projects, and other educational and community efforts, working to ensure that a self-advocate perspective is at the table. The slogan of the Self-Determination movement has long been “Nothing about us without us” and the CIDD is proud to become one of the few LEND programs in the country to add a faculty position that will ensure the inclusion of this critically important perspective.
Dr. Robert Emerson Receives NARSAD Young Investigator Award

Dr. Robert Emerson, a postdoctoral fellow at the CIDD, was recently awarded a NARSAD Young Investigator Award. This grant is funded through the Brain and Behavioral Research Foundation and provides support for the most promising young scientists conducting neurobiological research related to brain and behavioral disorders. Dr. Emerson's research will focus on understanding how the brain develops in infants at high risk for autism. For children who develop autism, early detection is critically important. The earlier they receive appropriate interventions, the more likely those interventions are to have a positive and lasting impact on their lives. One barrier to early detection and intervention is that the core symptoms of autism don't typically become apparent until the second year of life; however, brain differences often appear earlier than behavioral changes. Dr. Emerson's current research with Dr. Joseph Piven has leveraged this idea to identify patterns of brain activity in 6-month-old high-risk infants that can predict which infants will be diagnosed with autism at 24-months of age.

In the coming years, Dr. Emerson will focus his research efforts on verifying his early diagnostic method and explore the possibility of using infant neuroimaging to go beyond the prediction of diagnosis toward a prediction of an individual's future behavioral profile. He hopes his work will take the first step toward developing a clinically useful diagnostic test for detecting autism in infancy and help provide at-risk children with an individualized level of clinical care and intervention.

Dr. Adrienne Villagomez
Completes Certificate Program in Developmental Disabilities

CIDD psychology intern Adrienne Villagomez, PhD, completed the CIDD Certificate Program in Developmental Disabilities. To earn her certificate, Dr. Villagomez completed interdisciplinary coursework and attended campus seminars with a focus on developmental disabilities and conducted research related to developmental disabilities. Dr. Villagomez’s dedication to a career working with individuals with developmental disabilities was evidenced by her involvement in the North Carolina Postsecondary Education (PSE) Alliance, her work as a CIDD LEND (Leadership Education in Neurodevelopmental and Related Disabilities) trainee in Special Education, her participation as a CIDD practicum student, and her research focused on decisional capacity in individuals with fragile X syndrome.

Dr. Villagomez is pictured with the CIDD Certificate Program Director, Dr. Gabriel Dichter.

Eric Rubenstein Awarded 2016 Weatherstone Predoctoral Fellowship

Autism Speaks’ two-year Weatherstone Predoctoral Fellowships enable highly promising doctoral students to pursue autism research projects under the mentorship of the field’s leading scientists. Congratulations to UNC’s Eric Rubenstein, recipient of the 2016 Weatherstone Predoctoral Fellowship. Mr. Rubenstein will explore the association between autism symptoms in children diagnosed with the condition and autism-like behavioral traits in their parents (who don’t have autism). The goal is to better understand how and when inherited factors play a role in the development of autism and then use this information to tailor interventions that can best meet a child’s needs. The study also promises to deepen understanding of the inherited traits and biology of different subtypes of autism. Mr. Rubenstein’s fellowship mentors at UNC include epidemiologist Julie Daniels, public health statistician Amy Herring and CIDD psychologist Rebecca Edmondson Pretzel.
Shana Hall, PhD, received her doctorate from Duke University under the mentorship of Dr. David Rubin. Dr. Hall investigated the neural underpinnings of involuntary episodic memories in both non-disordered and posttraumatic stress disorder (PTSD) populations. Not only are involuntary memories a common but understudied phenomenon, but disruptive involuntary trauma memories are an important symptom of PTSD. This research has shown that in a non-disordered population the neural correlates of involuntary memories are remarkably similar to voluntary memories with the exception of decreased activity in frontal regions typically associated with effortful retrieval, suggesting that involuntary remembering does not require a special mechanism. However, in PTSD, neural activity associated with emotional involuntary memories peaks earlier than it does in a control population but it for non-emotional involuntary memories it peaks later. Dr. Hall joined the CIDD to work with Dr. Jessica Cohen to study bottom-up cognition in children with attention-deficit/hyperactivity disorder (ADHD). Dr. Hall will use graph theory methods to examine how disruptions in functional neural networks involved in bottom-up cognitive processes differ from disruptions in networks involved in top-down cognitive control processes and how these disruptions are differentially ameliorated by the administration of methylphenidate.

Candace Killian-Farrell, PhD, LCSW, is a clinical social worker with over 15 years of experience as a child/adolescent psychotherapist providing diagnostic assessment and evidence-based treatment to a range of children and adolescents, including those struggling with developmental disabilities. She received a master's in social work from Columbia University and her doctorate from the University of North Carolina at Chapel Hill. Killian-Farrell’s research has focused primarily on elucidating the risk processes associated with childhood stress and adolescent mental health outcomes in at risk populations. For her dissertation, she developed a transdisciplinary conceptual model examining stress response and neurodevelopmental outcomes as key mediating processes between childhood adversity and adolescent mental health. Dr. Killian-Farrell joined the CIDD T32 program to work with Dr. Aysenil Belger to study bottom-up cognition on elucidating the risk processes associated with childhood stress and adolescent mental health outcomes in at risk populations. For her dissertation, she developed a transdisciplinary conceptual model examining stress response and neurodevelopmental outcomes as key mediating processes between childhood adversity and adolescent mental health. Dr. Killian-Farrell joined the CIDD T32 program to work with Dr. Aysenil Belger to study bottom-up cognition and brain function, and negative symptoms in prodromal, at risk, and low risk adolescents. Her research aims to increase our understanding of the neurobiological underpinnings of stress regulation and negative symptoms in children with late-onset neurodevelopmental disorders. She hopes to begin to elucidate this link on a neurobiological level to better conceptualize these risk processes and eventually develop targeted interventions that may effectively address these detrimental developmental outcomes.

Alexander Tuttle, PhD, received his bachelor of science in psychology at Haverford College, where he first became interested in pain research working with Dr. Wendy Sternberg. After graduation, Tuttle spent two years studying cell cycle mechanisms in pancreatic beta cells with Dr. Jake Kushner at the Children’s Hospital of Philadelphia before deciding to return to the pain field. Working with Dr. Jeffrey S. Mogil, Tuttle recently obtained his doctorate in Psychology at McGill University. During his time at McGill, Dr. Tuttle designed a novel behavioral assay looking at prolonged social behaviors in mice as a way to study how pain and stress activation modulate social interaction. Currently, Dr. Tuttle is working with Dr. Mark Zylka to develop new, etiologically relevant assays in the pursuit of identifying new autism models. Specifically, Tuttle is interested in determining potential molecular, cellular and behavioral phenotypes in several high confidence autism gene mouse models. Tuttle is also interested in evaluating nociceptive and proprioceptive signaling in autism-like models; towards this end, he is developing several new behavioral assays to quantify novel social behaviors. By integrating autism genetics with animal models, Dr. Tuttle believes that common phenotypes between rodents and humans can be identified and further our understanding of autism spectrum disorders.
2016 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! Join us 6:30PM to 8:00PM in the CIDD Castelloe Conference Room 101.

Wednesday, October 12

Laura Hiruma, PhD
Psychology Postdoctoral Fellow

“Addressing Sleep Difficulties in Individuals with Developmental Disabilities”

Dr. Hiruma will provide an overview of the basics of sleep, sleep-related difficulties common among individuals with developmental disabilities, and sleep-related intervention strategies. Relevant resources for practitioners and families will also be reviewed.

Wednesday, November 9

Donna Carlson Yerby, MEd
Assistant Director: Services; Education - Carolina Institute for Developmental Disabilities

Deborah Zuver, MA, LMFT
Education Consultant - Carolina Institute for Developmental Disabilities

Ann Palmer, BA
Family Faculty - Carolina Institute for Developmental Disabilities
Parent Support Coordinator - TEACCH

“College Readiness for Students with ID and/or ASD: Strategies and Options”

This panel will discuss how students with intellectual/developmental disabilities, including those on the autism spectrum, can benefit from a college experience. Learn how students prepare to transition, access services, and advocate for themselves.

Wednesday, December 7

Mindy Govan, BS
TEACCH Certified Advanced Consultant, Adolescent/Adult Autism Specialist

“Teaching Relationships, Sexuality, and Safe Practices to Older Adolescents and Adults with ASD”

Ms. Govan will focus on how to assess what the individual with ASD is currently doing to meet their relationship and sexual needs, and how to work with them to clarify their goals and develop concrete strategies for addressing areas where they need assistance.

To RSVP or for more information, please contact:
Debbie B. Reinhartsen at (919) 966-4138 or Debbie.Reinhartsen@cidd.unc.edu
Yolandas Alston, who recently finished up her LEND training as a family fellow, has been an important part of the Durham County Department of Public Health’s Dental Clinic for children with autism spectrum disorder (ASD) and/or other intellectual developmental disabilities. Ms. Alston is also a parent of a child with ASD. The clinic’s goal is to ease the anxiety in children with sensory integration at an early age to provide more successful and routine visits in an environment that is welcoming and safe. Nutrition consultation is also provided for families to ensure the children are receiving a well-balanced diet and helpful ways to introduce new foods. This picture was taken at the 2nd Annual Autism Awareness Dental Clinic for Durham County residents. The event, initially started to serve patients annually, has now been expanded to a quarterly event. During her LEND training, Ms. Alston was mentored by LEND Family Faculty, Ann Palmer.

Photo credit: Durham County Department of Public Health

The neural correlates underlying social deficits in autism spectrum disorder (ASD) remains elusive, representing a fundamental gap in understanding the etiology of ASD. Shanna Resendez, a T32 postdoctoral research scholar in the laboratory of Dr. Garret Stuber, has been utilizing animal models to explore how a pivotal component of social brain circuitry, oxytocin neurons within the hypothalamus, encode socially relevant information. The overall aim of the research is to push forward the current understanding of neural mechanisms that mediate prosocial behaviors and that this research will aid in the development of therapeutic treatments for ASD. Ms. Resendez is pictured left at the Gordon Conference on ‘Fragile X and Autism-Related Disorders’ in Vermont where she presented the poster, Oxytocin regulation of socially motivated behaviors.

Poster Presentation at the Gordon Research Conference on Fragile X and Autism-Related Disorders

Connecting Autism Awareness & Oral Health Care

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Photo credit: Durham County Department of Public Health

Triangle Inclusive Employment Kickoff

CIDD affiliate Persever8 has teamed up with the Power of the Dream to create a new grass-roots movement for improving job opportunities for adults with autism and intellectual developmental disabilities (IDD). We invite friends and family members of people with autism/IDD to the kickoff event for "Triangle Inclusive Employment" (TIE) on October 17, 7:00 – 8:30pm, at the Frontier in RTP.

Through the TIE program, parents and family members of persons with autism will learn how to “Be an Employment Champion for Autism” at their work places. Parents will receive guidance and talking points about the advantages of employing this special population. Connecting families affected by autism will also create a network of Triangle professionals who are aware of the challenges their children face once they leave school. Understanding their options, available resources, and hurdles to employment will empower them to educate others, including their employers. The event is free but registration through Eventbrite (https://www.eventbrite.com/e/triangle-inclusive-employment-kick-off-tickets-27265403556) is encouraged.

Visit www.thepowerofthedream.org or http://www.persever8.org/ to learn more.
Congratulations to the CIDD All Stars, 4 time flag football champions and 2015-2016 basketball champs!

Team members front row: Lindsay Harris, Torri Hooker, Kristin Garrett, Jessica Hollando. Back row: Daniel Downing, Ernest Clemons, CJ Hooker, Darrious Massenberg. Not pictured: Brian Wrighten and Ryan Scotton

The UNC Autism Research Registry and SPARK team up at the Chipping in for Autism Golf Tournament to SPARK a movement in autism research. Pictured left to right: Corrie Waltson, Austin Ludwig, and Renee Clark.

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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Newsletter Editor—Keath Low