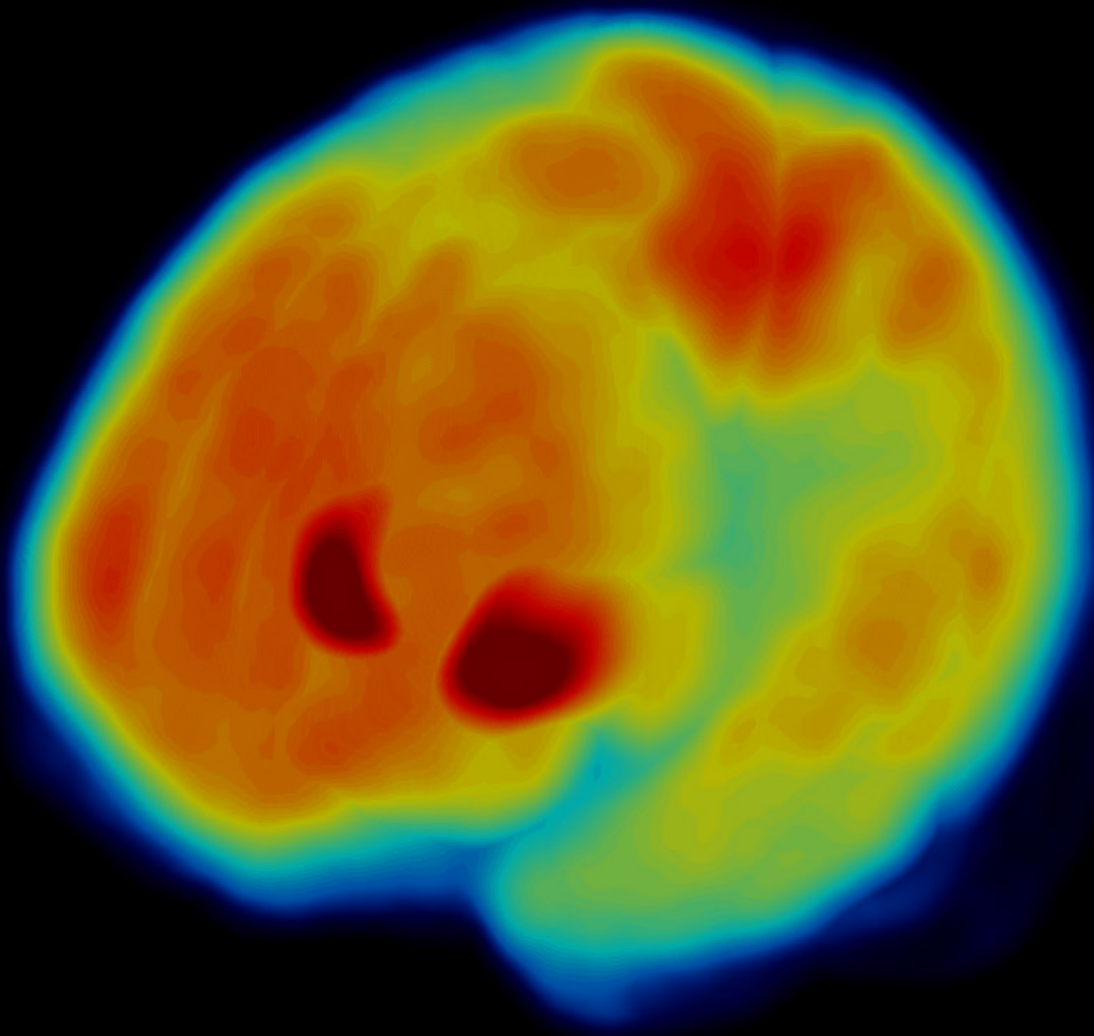


Intersections

Waisman Center, University of Wisconsin-Madison



Highlights Include

Inclusive Research

Autism & Aging

Transition to Adulthood

Autism & ADHD

COOL SCIENCE IMAGE WINNER

Matt Zammit, PhD, Waisman scientist in the Brain Imaging Core is a winner of UW-Madison's Cool Science Image Contest. Zammit's animation combines 15 years of study PET scans to show where protein plaques most often develop in individuals with Alzheimer's disease.

Intersections



Feathered Friends

This Sandhill crane family regularly visits the Waisman Center. You will often find them just outside the lobby doors. The crane couple even brought their chick to visit the center this year, as pictured above.

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Dear Waisman Community:

For more than 50 years the Waisman Center has been at the intersection of discovery and hope – forging new paths in research and clinical care. One of our strengths is the diversity and breadth of the research, service, training, and outreach efforts that take place under our roof and extend out to the community. The work is transformative in the advances we make and in the care that we provide for individuals and families impacted by intellectual and developmental disabilities and neurodegenerative diseases.

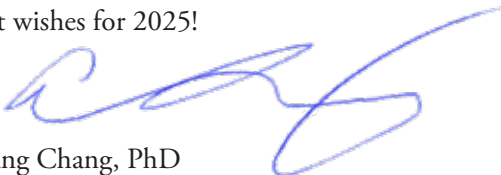
I am delighted to highlight a selection of this year’s achievements and advances within the pages of our impact report. As you flip through the pages, there is so much promise for future treatments and interventions as we broaden our understanding of aging in autism and Down syndrome, make earlier diagnosis of cerebral palsy, and include individuals with lived experience as advisors and co-researchers. Standing together on the foundation we jointly built, we are excited about all kinds of opportunities on the horizon and look to accomplish even more in the years to come.



You have been an invaluable partner to our success up to now. Your new contributions will sustain the center’s mission and enable us to continue to improve the lives of individuals and families with IDD through research, service, training, and outreach activities.

On behalf of the entire Waisman community, thank YOU.

Best wishes for 2025!


Qiang Chang, PhD
Director, Waisman Center
Professor of Medical Genetics and Neurology

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A Family's Commitment to Down Syndrome Research at the Waisman Center Spans Four Decades and Counting

By Emily Leclerc



(From left to right) Sigán Hartley, Heather Huismann, Brad Christian, and Renee Makuch at the Friends of the Waisman Center Awards Reception where Huismann was honored for her volunteer efforts.

When Heather Huismann was in middle school she called the police on her teacher. “He was not teaching me very well,” Heather says. So, she did what her parents taught her to do when there was a problem – stand up for herself. “She called 911 to get her teacher fired,” Larry Huismann, Heather’s father, recalls fondly. Heather has no hesitation when it comes to speaking out and getting things done – a very take-the-bull-by-the-horns approach, as she says. Her fiery spirit and fierce determination have served her well in the decades since middle school, especially when it comes to handling the challenges that can come with living with Down syndrome. That determination and her family’s unwavering support are part of why Heather has been participating in research at the Waisman Center for more than four decades.

“The Huismanns are a family that have been with us for a very long time. It has been so meaningful to get to see Heather and her family over the years, to hear their stories, and hear about their successes and challenges over time,” **Sigán Hartley, PhD**, Waisman investigator and 100 Women Distinguished Chair in Human Ecology says. “The importance of having families stay with us for long periods of time is invaluable to us and to the science. The Huismanns have been a delight to work with.”

Down syndrome (DS) or trisomy 21 is a genetic disorder where an individual is born with an extra 21st chromosome. It is the most common chromosomal condition with an estimated one in every 775 children born in the United States receiving a diagnosis, according to the Centers for Disease Control and Prevention. Down



syndrome is typically accompanied by distinct facial features, developmental delays, intellectual disability, and is associated with increased risk of several different health conditions such as heart disease and Alzheimer's disease. Even with these commonalities, every person with Down syndrome is unique and their experiences vary greatly.

After Heather was born, her mother Pam quickly started looking around for resources and supports for her daughter. As a mammographer and CT tech in a clinic in Sun Prairie, WI, Pam already had a working knowledge of Down syndrome and potential support options in Madison. She knew she wanted to enroll her daughter in research opportunities in order to learn more about her and the Down syndrome community at large. "I don't know how she came up with the Waisman Center but I am glad she did," Larry says. "It has been a long relationship and it has been just wonderful."

Heather has been participating in research at the Waisman Center since she was 3 years old. Her first study was with former Waisman investigator Len Abbeduto, PhD, currently a psychologist and professor of psychiatry and behavioral sciences at the University of California – Davis, and retired co-director of the MIND Institute Intellectual and Developmental Disabilities Research Center. That first study was 42 years ago. In her almost four and a half decades being involved at the Waisman Center, Heather has participated in many different studies for many different Waisman investigators all with the intent of broadening the collective understanding of Down syndrome.

When asked why she has remained so dedicated to being a part of research, Heather credits her parents' loving influence with continuing to push her to take part in something bigger than herself. "It was mainly because of the love and the support and the guidance I got from my mom and dad. That and I am just trying to show people who I really am," Heather says. Pam and Larry have stood solidly behind Heather ever since she was little and strove to give her every opportunity possible.

"These advances would not be possible without the steadfast commitment of our families to stick with us over the years and Heather is a shining example of the amazing people that work with us."

-Brad Christian

At the moment, Heather is participating in the Alzheimer's Biomarkers Consortium – Down Syndrome (ABC-DS) study co-led by Waisman investigators **Brad Christian, PhD**, professor of medical physics and psychiatry and Sigan Hartley. The ABC-DS study is a large longitudinal study focused on trying to identify early biomarkers of Alzheimer's disease in order to help inform clinical trials and improve the quality of life for individuals with Down syndrome and the general population. People with Down syndrome are at high risk for an Alzheimer's diagnosis with an estimated 90% of individuals developing

clinical symptoms of the disease in their lifetime. A large part of why Heather decided to join this study is she lost her mother several years ago to Alzheimer's disease.

Sadly, Pam Huisman passed away in 2017 but Heather knows

that if she were still here that she wouldn't want Heather to stop her involvement with Waisman. "If my mom were here right now, she would have told me to continue with the influence and impact that I've had with the Waisman Center," Heather says. "My mom was the most influential person in my life." So, she continues to volunteer her time to help Waisman's researchers expand their understanding of Down syndrome.

Larry has also stepped in to be Heather's research partner after his wife's passing, accompanying her to many of her research visits and supporting her as needed. "Pam left some large shoes to fill but I am doing my best," Larry says. He is a steadfast advocate for Heather and always ready with a quick joke to entertain the research teams. It is abundantly clear that he not only deeply loves his daughter but is also incredibly proud of the woman she has become and the influence she has had on those around her. The two of them make an excellent team capable of just about anything.

"Larry is a very supportive father and has beautifully filled the role of Heather's study partner after his wife passed away. Both are a joy to work with on our testing days," **Renee Makuch**, study coordinator for ABC-DS, says. "Larry always arrives with a few new jokes which keeps us on our toes and leaves us all with smiles. Partnerships with families like theirs are imperative as we strive to improve the quality of life for individuals with Down syndrome as they age. Our research group is



Heather and her father Larry at the Waisman Center

Larry so much. It hasn't just been the researcher's that have benefited. Being a part of the different studies over the years and interacting with experts in the field have given the Huisman's a better understanding of resources and tips, tricks, and tools for working through challenges. "One of the things Waisman has really done for us as a family is allowed us to recognize opportunities. If that person can do that, why can't we?" Larry says. "And if we set the bar high then there's no reason this other person can't come up and enjoy accomplishing those opportunities as well."

Research into conditions like Down syndrome is critical for improving not only our understanding of the condition as a whole but also treatments, therapy, and their overall quality of life. And without people and families like Heather, Larry, and Pam, this type of research would not be possible. "They've gone through many different years of a person's life, from infants to adults. Their experiences can help us through something and maybe they will learn from our experiences," Larry says. "It is a true pay it forward thing that I feel is incredibly important."

With her dad and her two sisters, Missy and Erin, as her advocates, Heather continues to volunteer her time to help the Waisman Center advance knowledge of Down syndrome. "Everything that my mom and dad did for me," Heather says. "I still have that in me. And I want to be there at the Waisman Center." ■

tremendously thankful for their continued support and contribution to our study."

Both Brad and Sigan always look forward to Heather and Larry coming in for research visits. Not just because they are a joy to be around but also because of their appreciation for the research they are participating in. They are both deeply grateful to Heather and her family for their dedication over the years.

"The contributions made by Heather and her family in advancing Down syndrome research at the Waisman Center have been extraordinary! Heather is very busy with family, work, and social activities but she is always incredibly giving with her time to participate in our research studies," Brad says. "Remarkable advances are being made in understanding the changes in the brain related to Alzheimer's disease in people with Down syndrome and our study is helping to chart the course for future prevention and treatment trials. These advances would not be possible without the steadfast commitment of our families to stick with us over the years and Heather is a shining example of the amazing people that work with us."

Participating in research has also given Heather and

Alzheimer Biomarkers Consortium – Down Syndrome (ABC-DS)

The Alzheimer Biomarkers Consortium-Down Syndrome (ABC-DS) is a longitudinal study funded by the National Institute on Aging (NIA), the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD), and the National Institutes of Health (NIH) INCLUDE (INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndrome) project.

The goal of ABC-DS is to follow a cohort of adults with Down syndrome over time to identify early biomarkers that may herald the onset of Alzheimer's disease. The investigators hope that these biomarkers can be useful to inform clinical trials and improve the quality of life in people with Down syndrome and for the general population. The Waisman Center is part of this multi-site study across the United States and at University of Cambridge in England.



Golfers at the Friends of the Waisman Center golf benefit

Special Events

FRIENDS OF THE WAISMAN CENTER SPRING BENEFIT

June 1

Spring benefit donors enjoyed a catered meal by Chez Vous and wine donated by General Beverage and Phillips Wine Company delivered to their home by Friends board members.

MORSE SCHOLARS REUNION

June 13-14

Current and former Morse Scholars converged in Madison for a reunion and meeting with society founder and namesake, Dr. Richard Morse and Larry Connor.

ASSOCIATION OF UNIVERSITY CENTERS FOR EXCELLENCE IN DEVELOPMENTAL DISABILITIES (AUCD) BOARD RETREAT

June 25

The national AUCD board of directors traveled to Madison and visited the Waisman Center as part of their board retreat. The 16-member board is made up of professionals, individuals with disabilities and family members who are elected to serve a three-year term.

FRIENDS OF THE WAISMAN CENTER GOLF BENEFIT

August 5

24 teams of golfers played FORE a good cause at the Friends of the Waisman Center Golf Benefit held at Maple Bluff Country Club. Proceeds from this event help fund a broad range of Waisman activities and programs.

MAGNUS HIGH PERFORMANCE NEUROIMAGING SYMPOSIUM

September 17

The Waisman Center hosted the High-Performance Neuroimaging Symposium, which brought researchers from all over the country to share and learn about the MAGNUS MRI, a new, cutting-edge MRI technology the Waisman Center Brain Imaging Core acquired in 2023.

WAISMAN WHIRL RUN, WALK & ROLL FOR ALL ABILITIES

September 29

More than 300 people walked, ran, and rolled to raise awareness and funds in support of the Waisman Center. Hosted by the Friends of the Waisman Center.

IDDRC NETWORK DIRECTORS MEETING

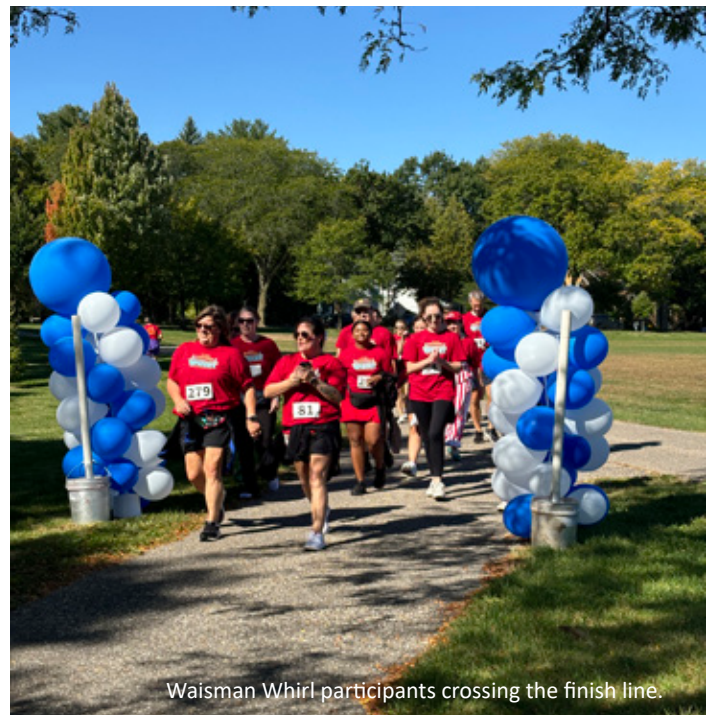
October 9-11

The Waisman Center hosted directors and delegates from the network of Intellectual and Developmental Disabilities Research Centers (IDDRC) for their annual meeting. Presentations focused on advances in research on intellectual and developmental disabilities, including the use of artificial intelligence, and the inclusion of individuals with disabilities on research teams.

DISABILITIES RESOURCE FAIR FOR HISPANIC FAMILIES

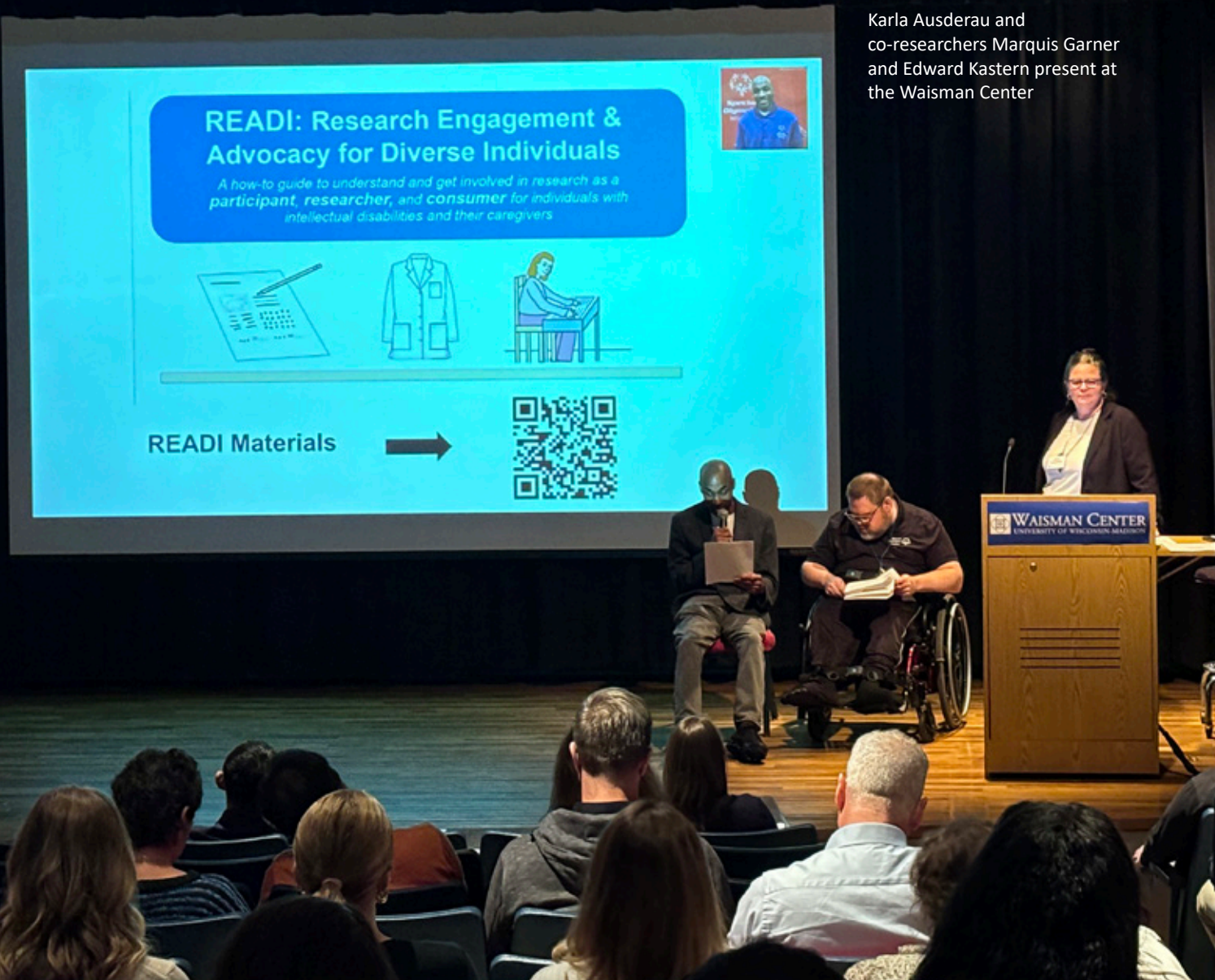
October 23

The Waisman Center held its second annual resource fair for Hispanic families of children and adolescents with intellectual and developmental disabilities at Aldo Leopold Elementary School. More than 30 community partners and Waisman Center programs provided information in Spanish about disability resources, services, and supports.



Waisman Whirl participants crossing the finish line.

Karla Ausderau and co-researchers Marquis Garner and Edward Kastern present at the Waisman Center



Advising Research through the Lens of Lived Experience: How Individuals with Disabilities and their Families are Shaping Research

By Charlene N. Rivera-Bonet

Each individual interviewed for this story stated their preference between person-first or identity-first language. The language used reflects the preference of each person. Researchers interviewed use identity-first language in alignment with the preferences of many of those in the autistic community.

Who is considered an expert in autism? Or cerebral palsy? Or Down syndrome? The word expert is often used to describe someone who has studied a topic extensively, whether through research, through school, or

practice. But what about people with lived experiences?

The lived experiences of individuals with intellectual and developmental disabilities (IDD) offer a unique and deeper perspective, which is valuable when considering the research questions that are of most importance to themselves and their community. The ups and downs, what works and what doesn't, what they need and what they don't. Many investigators at the Waisman Center are turning to individuals with lived experiences as advisory board members to provide expertise and perspective

that researchers may not have.

Advisory board members with a lived experience – whether that is a person with an IDD or a family member – are working to make IDD-related research more meaningful, accessible, and representative of the views, priorities, and experiences of their community. These individuals might traditionally be considered participants in a research study, but now work side-by-side with researchers as collaborators.

“NOTHING ABOUT US WITHOUT US”

“The adage, ‘nothing about us without us’ is a really important thing to think about as an autism researcher and as a disability researcher more broadly,” says **Lauren Bishop, PhD**, Waisman investigator and associate professor in the Sandra Rosenbaum School of Social Work. The disability community has been historically left out of conversations about research, Bishop says. “So, making sure that you’re partnering with folks who are members of the community throughout the research process is really important to maintain the integrity of what you’re doing, and to produce research that is impactful and meets community needs.”

Karla Ausderau, PhD, professor of kinesiology, Occupational Therapy Program, formed her first advisory board in 2016 to get feedback on an intervention her lab developed for improving feeding in autistic children. “We had parents who were an advisory board and they gave us feedback on the intervention. What was feasible, what might be burdensome, what types of content we should include in the parent training manuals and how to best deliver that content,” Ausderau says.

Katy Ptaszynski offers her perspective as a parent on behalf of her son Xander, a 10-year-old autistic boy with ADHD. She is part of one of the advisory boards from the lab of **Brittany Travers, PhD**, associate professor of kinesiology. For Ptaszynski, making the decision to participate on the board was tough, because she considers herself to be neurotypical, and is representing her son, who is not. But she also thinks it’s important to represent people from all ages. “So as a parent, I’m kind of in that role right now. I want to be able to speak on behalf of my son, but at some point, I’d really like him to be able to advocate for himself,” Ptaszynski says.

Travers has two advisory boards for her research projects on autism and motor performance. “They’ve offered up paradigm-shifting information and interpretations of our data that have really helped us plan future experiments and research,” Travers says.

Carlos Schaut is an autistic man who has been using a board to spell as his preferred method of communication for 21 years. He visited the Waisman Center Clinics when he was younger, but now offers his expertise as an autistic speller to Travers’ research. “I was invited [onto the board], and so I wanted to offer my input as a speller,” Schaut says. “It has been important to add my words to the discussion and offer input. I think it is very important to include

non-speakers because we have been left out for so long,” he adds.

THE PROCESS OF ESTABLISHING A BOARD

For researchers, it is important to have individuals from different backgrounds, levels of need, and abilities in order to make their research more accessible and representative of the community. “It’s been very interesting to me to just see some of the differences between the board members and how they talk about autism and what’s on their mind in terms of their needs,” says Travers about her advisory board dedicated to making her lab more accessible to the Spanish-speaking community.

“We’ve worked really hard to make sure that our team is diverse, and that we have members from diverse racial and ethnic groups and regions, to make sure that we are capturing different perspectives,” says **Sigan Hartley, PhD**, professor and 100 Women Distinguished Chair in Human Ecology. Hartley studies factors that shape healthy development in people with neurodevelopmental disorders such as Down syndrome.

Ultimately, the goal is to produce research that is meaningful to the community. Because of this, the community board’s involvement in research projects begins even before the project takes shape. “The key part, from my perspective, is that those people are brought in from the time that you are thinking about the project, and drafting the proposal. Not as an afterthought,” says **Rebecca Alper, PhD**, assistant professor of communication sciences and disorders. Alper’s work focuses on improving language, academic and health outcomes for at-risk children through family-centered early language and literacy intervention.

The process of creating an advisory board requires a lot of thought and intentionality. “Thinking about what is the purpose? How are they going to help move this [research] forward? And what is the mechanism for implementing change based on the advisory board input? And I think that starts at the very beginning,” Ausderau says.

Once the boards are formed and expectations for each side are clear, they meet to discuss research. These meetings typically include the researcher presenting project ideas first, and then having discussions with the board. “When Brittany [Travers] or **Emily [Skaletski]**, graduate student in Travers’ lab] explain the research, sometimes it’s a little daunting, it’s a little difficult to understand, but they definitely break it down in a way that’s translatable,” Ptaszynski says. After that, “we comment as we feel led,” Schaut says.

The next steps are vital: listening and implementing. “I think that would be very frustrating, if you bring a community partner in to give feedback, but there was actually no room to change anything. That would be literally just lip service. So really going into it as a partnership. Not just a box to check off on your list,” Alper says.

This was one of Travers’ concerns when first forming

her board. “I was really nervous about it at first. The research that I do is my obsession and it felt really scary to open that up to another group of individuals,” Travers says. “And at the same time, I just have such a respect for and passion for working with autistic individuals. I didn’t want to disappoint the community advisory board if there were things that they recommended that we couldn’t implement.” However, this didn’t stop her from forming a board, and she is reaping the fruits of her choice. “And I will say our community advisory board is probably the best thing that’s ever happened to our lab in terms of the research. It’s provided us with really amazing insights,” Travers says.

When Hartley and her team were designing a study, one of the things that families brought up was providing research participants with more immediate and meaningful information stemming from their participation. They came up with the idea of a “health report card”, containing a summary of the child’s cognitive, health, and behavioral performance based on the measures collected from the study. In addition to sending this “health report card”, families suggested sending a list of resources so that the family could seek out more information or services if appropriate. This helped make Hartley’s research more meaningful to the community.

BEING PART OF THE RESEARCH TEAM

Another way individuals with IDD bring their expertise to the research team is by becoming a co-researcher. In this position, they help with the details of the research process, including data collection, analysis, and interpretation.

Amy Frechette is a co-researcher in one of Bishop’s studies that looks at using machine learning algorithms to

establish autism mortality risks indices. Co-researchers are community members that are embedded within research teams working side-by-side as collaborators. Frechette was diagnosed with autism (Asperger’s at the time) in her late 20s. She has become an avid advocate and leader, serving as a governor appointed self-advocate to the New Hampshire Council on Autism Spectrum Disorders, and providing support to the Leadership Education in Neurodevelopmental and Related Disabilities Program (LEND) in her state. For Frechette it is important for those with a disability to be included in research. “It goes with a lot of different work that I’ve done over the years. And it’s a way for me to help move our country’s agenda a little further ahead, because they always tell me that people with disabilities are an underserved, underrepresented population,” Frechette says. So, when she got the invitation to become a co-researcher with Bishop, she thought “Oh, yeah, well, this is a nice opportunity. I should do it.”

Part of Ausderau’s research is thinking about how to build capacity for people with IDD and their care partners to actively engage in research. In collaboration with the Special Olympics and other universities, Ausderau developed training modules that help individuals with IDD learn what research is, why their involvement is important, and how to be involved. To her, it is not only about bringing them to the table, but also providing them with the tools to give them the confidence to contribute to the conversations. She has also helped develop an ethics training specific for individuals with IDD in research teams. “How can we think about regulations at an institutional level to facilitate and make things more inclusive?” Ausderau says. Part of this work, is also

training neurotypical researchers to work with individuals with IDD and increasing university research accessibility for individuals with IDD as Ausderau and community members have addressed.

Co-researchers in Ausderau’s lab are involved in all aspects of a project. “They help to think about what questions we’re asking. They will help us with recruitment, if they’re part of the IRB (Institutional Review Boards). They help us think about better ways to collect our data. They will help us with analysis and interpret the results we’re finding,” Ausderau says.

An example of this was an interview study they were doing with individuals with IDD. Team members suggested that they needed a more creative methodology because interviews and voice communication wasn’t



The Ptaszynski Family



Karla Ausderau and co-researchers Marquis Garner and Edward Kastern

about the work that we do when we involve community partners, is it shortens the timeframe for getting research implemented and being taken out by our community members. Because if team members are on board from the very beginning, it really helps us know and have a better pathway for the types of research we're doing and it helps us think about the implications of what we're doing and then how to disseminate it." ■

always the best option for everyone. So, they adapted photovoice as an alternative methodology, using pictures to guide their meetings or discussions and findings. "And so that was a really instrumental time for us thinking about how can we use another methodology that's already very valid in the literature and then adapt to meet the needs of the population we are working with," Ausderau says.

BENEFITS OF PARTICIPATION IN RESEARCH TEAMS

Research from Ausderau's lab has found that including individuals with IDD as co-researchers is both beneficial and feasible. Co-researchers find their work to be meaningful, and helpful for skill development.

"One of the reasons I also wanted to join is I don't know a lot about the research community, and it was just actually a great learning experience," says Ptaszynski about her participation in Travers' advisory board. "Some of the members of the board said that it was actually helping them learn to trust science more and understand scientific perspectives," Travers says.

Both advisory board members and co-researchers receive paid compensation for their work.

IMPROVED RESEARCH OUTCOMES BECAUSE OF INCLUSION

Alper, Ausderau, Bishop, Hartley, and Travers all agree that their boards and co-researchers have been highly beneficial to their research. "I think we feel more confident that what we're doing actually is going to meet a community need," Ausderau says. "I think one of the most important things



SELF-ADVOCATES BRING RESEARCH TO THE COMMUNITY

Maria Nogueron and Danny Chafetz (above) are part of the outreach team for the Alzheimer's Biomarkers Consortium – Down Syndrome (ABC-DS) in the lab of Sigan Hartley, PhD. Chafetz, a man with Down syndrome, and Nogueron, a mom of a woman with Down syndrome, bring their lived experiences and expertise in Down syndrome to recruit individuals to participate in research to advance understanding of Alzheimer's in Down syndrome. They also share the science learned from studies in the lab with the community.

Autism through Midlife and into Older Age

New study helps understand changes in autism as individuals age

By Charlene N. Rivera-Bonet



Autistic adults may face changes in their autism symptoms, behavioral functioning, and health as they age, with some improving and some worsening, a new study shows. This insight may help autistic individuals and their families prepare and plan for the future.

Autism is heterogeneous across the autistic population, but it is also not static across the lifespan of a single person, the Waisman Center study published in the *Journal of Neurodevelopmental Disorders* shows.

Adulthood is the longest stage in the life course, but until recently, the bulk of autism research was focused on early childhood, and research has not kept up with the changing needs of autistic individuals as they age.

The study, from the **Lifespan Family Research Lab** at the Waisman Center, aimed to identify points during the life course of autistic individuals when vulnerabilities may be increasing, or decreasing, in order to better understand how to best meet the needs of this increasingly large population.

The researchers measured age-related changes in autism symptoms, behavioral functioning, and health prospectively

over 22 years, extending from adolescence through midlife and into the early years of old age. The data comes from an ongoing longitudinal study of families of autistic adolescents and adults that began in 1998 and includes 406 autistic individuals who ranged in age from 10 to 52 at the start of the study.

Specifically, they looked into what improved and what worsened, and whether the change was linear or not. They also evaluated whether these changes differed between those who have intellectual disability (ID) and those who do not.

“[The results] show yet again the heterogeneity of autism,” says **Leann DaWalt, PhD**, director of the University Center for Excellence in Developmental Disabilities (UCEDD) and co-principal investigator of the Lifespan Family Research Lab.

They first compared individuals with autism with ID to those with autism but no ID. “One of the things we have done was look at possible different paths between those who had intellectual disability and those who don’t,” says **Jinkuk Hong, PhD**, research scientist in the lab and first author of the study.

Results showed that for autistic adults with an ID, overall there was greater impairments in the ability to engage in social interactions and communication, less independence in daily living skills, higher levels of behavioral problems, less social time spent, and a higher number of non-psychotropic medications prescribed compared to autistic adults without an ID. How symptoms, behavior, and health changed throughout the life course also differed between autistic individuals with and without ID.

For both groups, some areas improved with age, some worsened, and some varied throughout different periods of their life. “Adulthood is not a static period for autistic adults. It’s not a static period for any of us. But there hasn’t been this research in the field of autism to trace the ups and downs of adulthood, when they occur, and what they are,” says **Marsha Mailick, PhD**, emeritus vice chancellor for research and graduate education and co-principal investigator of the lab.

The first pattern they found was a significant improvement over the adolescent and adult years for impairments in social reciprocity for those without an intellectual disability and impairments in verbal

communication regardless of ID status. Both of these became less severe as autistic individuals aged.

The second pattern was of significant worsening over the adolescent and adult years for all indicators of health. Ratings of health, numbers of prescribed medications, and physical health problems all worsened with advanced age. Although this may also be true for the general population, some unknowns remain such

as whether the worsening of these health symptoms begins at an earlier age for autistic individuals.

Other patterns identified were not as straightforward. Activities of daily living, behavioral problems, repetitive behaviors and socializing with friends and relatives all improved during adolescence into adulthood, leveled off, and then worsened in midlife and beyond.

“It’s a complicated story being revealed. And these were just some of the indicators of aging, but the complexity is important, and at some level, it wasn’t surprising,” Mailick says. “Because for all of us in the general population, aging is complex. It’s multifactorial. The timing of aging in one domain doesn’t predict the timing of aging in another. But again, we don’t have this information about autism.”

Currently, autistic individuals can access more services while they are in high school than those they can access during adulthood. Midlife seems to come with periods of growth and decline for autistic individuals, and this research points to the need for the services of autistic adults to change with advancing age.

This research also aims to help autistic adults and their families better plan for their future as they age. “We should be encouraging autistic adults and their family members not to think that the changes end as soon as they’ve made the transition from high school into the early 20s,” DaWalt says. “We should be thinking about all of the life course transitions that are going to happen in the life of autistic adults. We should be anticipating change.” ■

This study was supported by grants from the National Institute on Aging (R01AG08768), the National Institute of Mental Health (R01 MH121438), Autism Speaks (#7724) and the Waisman Center’s IDDRC core grant (P50HD105353).

“We should be thinking about all of the life course transitions that are going to happen in the life of autistic adults. We should be anticipating change.”

-Leann DaWalt

Leading by Example: Brooke Mortensen, NP

The Wisconsin Leadership Education in Neurodevelopmental and Related Disabilities (WI LEND) program trains the next generation of leaders to improve the health and well-being of individuals with disabilities

Brooke Mortensen, NP, always wanted to be a nurse. “I’ve always enjoyed science but I also love human interaction and connection. I feel that nursing really embodies those two spirits equally,” Mortensen says. Today, she is a pediatric nurse practitioner at UW Health American Family Children’s Hospital in the Complex Care Program. The UW Health Complex Care Program is designed for children with medical complexity to help manage their care and support the family through the various stressors they face. And without the WI LEND Program, Mortensen says she wouldn’t be where she is today.

Several years ago, Mortensen was working as an ER nurse and decided to go back to school to become a nurse practitioner. At the time, she was also working with a few patients in their homes, one of whom was a boy with paraplegia. “He really opened my eyes to the challenges that families face. He lives in Madison, one of the most well-resourced areas in the state, and yet he and his family were constantly running into barriers with getting the supports he needs,” Mortensen says. “He helped me realize that there was a real need to connect the dots for these families.” He is the one who inspired Mortensen to specialize in working with children with disabilities.

As she went through school, she realized that she needed more formal education and training working with people with disabilities. After speaking with several people, Mortensen was recommended to the WI LEND program. “I was told it would give me a better, more holistic view of working with people with disabilities, not just from a nursing perspective but from a multidisciplinary perspective,” Mortensen says. One application and several interviews later, Mortensen was accepted into the program at the Waisman Center.

“LEND was amazing. It changed my life. Truly it did,” Mortensen says. LEND’s variety of methods for imparting information, hands-on training, and clinical and family experiences, all came together to give Mortensen a comprehensive understanding of what it means to live with a disability and how she can provide the best and most respectful care possible.

Then it was through connections she made during LEND that

Mortensen first learned about the Pediatric Complex Care Program at UW Health. Without the training she received through WI LEND, she says she would have been unlikely to get her current job. “I would not be where I am today,” Mortensen says.

When asked about her biggest takeaways from the training program, Mortensen has two answers: remembering how resilient families are, and the importance of language. “One of the mantras I developed from LEND is that families are navigating a world that is not built for their child, and often exceeding expectations while doing it,” Mortensen says. Her second is all about language.

The language around disability has evolved greatly over the last several decades and continues to change rapidly. She stresses the importance of understanding how an individual and their family wants to be represented because it shows respect for their unique experience. “All families have their own different perspectives on how they view disability. And it is our job as healthcare providers to honor wherever they’re at in their journey and do what makes sense for them at the time,” Mortensen says. “We can’t come in with a preconceived agenda or notion of what living with a specific diagnosis looks like or means to someone.”

Mortensen is grateful for what LEND provided her, knowing she would be in an entirely different place without the program. She loves where she is and the families she gets to work with every day. Being a part of their team is a special honor for her.

“I love when families and children come in for an appointment and show us a new skill or milestone they’ve reached that they were told they would never have. I had a six-year-old a few weeks ago who was taking steps in the exam room and her family was told she would never be able to walk. Those are beautiful moments to witness,” Mortensen says. “Those experiences remind me all the time that yes, I have education in how a disability may present, and it’s my job to offer evidence-based diagnostics and treatments, but the child and family are the leaders. We’re there to walk alongside them and share our knowledge, not to dictate how their journey is going to look.”



Wisconsin LEND



2024-2025 WI LEND Cohort

WI LEND or the Wisconsin Leadership Education in Neurodevelopmental and Related Disabilities program is a training program based at the Waisman Center that provides interdisciplinary leadership training for individuals across disciplines to improve systems of care and access to resources and services for children with neurodevelopmental and related disabilities and their families. The program covers an entire academic year.

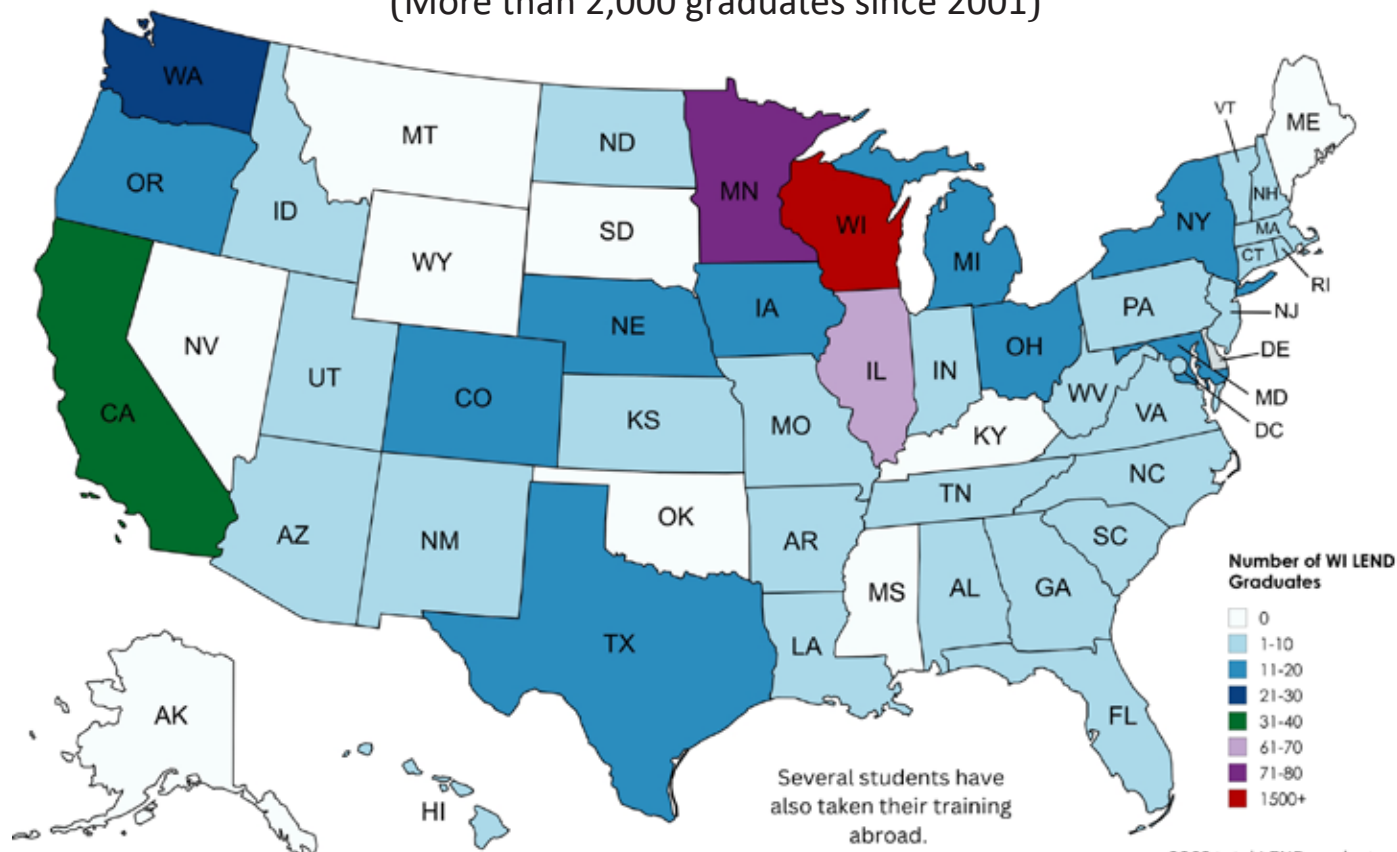
Training involves lectures and seminars, interdisciplinary small group work, working in the community with families, shadowing in a clinic, completing a research project, family mentor experience,

leadership development workshops, and mentoring. LEND graduates leave the program with a broad spectrum of knowledge and experiences that enable them to be more effective advocates for the disability community.

The cohort of LEND trainees includes a range of disciplines to underscore the importance of interdisciplinary perspectives and expose the cohort to the variety of disciplines that work with individuals with disabilities. Each cohort includes a parent or family member of an individual with a disability and a self-advocate – an individual with a disability.

Distribution of WI LEND Students after Graduation

(More than 2,000 graduates since 2001)



Brazil	1
Singapore	1
South Korea	2
Canada	3

2082 total LEND graduates





CHANGE AHEAD:

Creating a Road Map to Help Families Navigate the Transition to Adulthood for Individuals with an Intellectual and Developmental Disability

By Charlene N. Rivera-Bonet

Luis “Lui” Montoto’s dream is to open up a food truck where fast food doesn’t sacrifice being healthy, filling, and delicious. He loves being in the kitchen and cooking for his family. He is attempting to get his degree in culinary arts at Madison Area Technical College to become a sous-chef, but has run into a few major roadblocks: not being able to pass a requisite class and a cancer diagnosis. Some aspects of Lui’s transition into adulthood as a man with autism have been bumpy, unusual, and uphill to say the least. And yet, he has pushed forth with the help of one thing that keeps him steady: his family.

The road to adulthood can be rough terrain. It consists of uncharted territory and many changes. This can be even more challenging for individuals with intellectual and developmental disabilities (IDD), who may often rely heavily on resources, supports, and routines that can change or end once high school is over or once they are 18.

“There are a number of barriers and challenges that youth with disabilities may have as they look at transition [into adulthood],” says **Tim Markle, MA, MA/CS**, director of the Wisconsin Youth Health Transitions Initiative (YHTI) at the Waisman Center.

Understanding the multi-faceted and complex process of transition, YHTI, along with other programs at the Waisman Center, work to help make this trek smoother by providing resources, support, and guidance for individuals with IDD and their families, and training service providers to support transition in a holistic way.

LIFE AFTER HIGH SCHOOL

For adolescents with IDD, the transition out of high school comes with many challenges, from changes in routine, to entering new systems in which supports are not as involved. “Where I had trouble most was in the beginning parts of college,” Lui recalls.

For Lui, the transition into college was a big challenge because he no longer had his usual systems of support in place like his paraprofessionals in high school. “I had a bit of a learning curve in order to adjust from going from an easier setting to a more harder, higher-paced and stricter system when it came to my grade scale,” Lui says. “Especially for college, because I wasn’t actually brought up on that specific system for a while, so it made it a bit difficult, and plus, with my social skills, it makes it pretty hard for me to make connections with people as well.” He ran into the situation of not being able to pass a requisite class to get into the culinary program. Which delayed him for more than two years.

Even though the school does make accommodations for him, and tutoring is available, there isn’t anyone helping him make sure those



Blake Edison in front of some of his art.

accommodations are met and working, and sometimes it can be a challenge for him to know how and who to ask for help.

FINDING THE RIGHT MIX OF NEW ROUTINES

For Blake Edison, an easygoing, energetic 31-year-old artist with autism, the biggest challenge in the transition to adulthood was the change in the routine he had found great comfort in. When he was done with high school his parents Sheri and Glenn were faced with the question of what was next for Blake. “When the schedule of school has pretty much filled your young person’s day from 7:30 a.m. to oftentimes 3:30 or 4:00 p.m. each day, then you have to stand back and say, ‘How do I recreate a life for my loved one, whether it’s academic, vocational, social, or all of the above?’” Sheri says. When Blake was in 10th grade they started thinking about how to help him create a life that a typically-developing young person would create on their own.

For his parents, the biggest challenge was refusing to be limited by what people said was available for Blake. “We actually took a much broader approach and said, ‘What kind of life would give Blake a quality of life given who he is as a person, what he enjoys, what his talents are, and so forth,’” Sheri says. That meant that they weren’t always looking at programs designed for people with disabilities.

“We feel like we have discovered a bit of the secret sauce

of what makes a quality life [for him],” Sheri says. Blake works part time five days a week, goes to the ArtWorking studio twice a week, and works out with a personal trainer a few times a week.

HEALTHCARE AND GUARDIANSHIP

Before Lui graduated high school, Lupita and Luis Montoto, Lui’s parents, filled out paperwork to keep guardianship of Lui even after he became an adult, allowing them to make important legal decisions on his behalf. This isn’t always needed for individuals with IDD. Only when they are not able to make legal decisions by themselves. This was a huge help when Lui received his cancer diagnosis of T-cell lymphoblastic lymphoma/leukemia in 2021. “I don’t

think I have had any issues trying to access treatments or anything for Lui, because thank God, I got the guardianship,” Lupita says. Even during the COVID-19 pandemic Lupita was able to attend every appointment and treatment session with her son. “I was always with him every single time,” she adds.

Despite their ability to make decisions on his behalf, Lupita and Luis have always consulted Lui on all decisions regarding his healthcare. At times, Lui decided he was done with a specific treatment, and preferred a different course of action, and they respected those decisions. “Because at the end of the day, he can decide and has the last say,”

Lupita says.

“Healthcare is a huge area of transition that usually is not thought about,” Tim says. When children with IDD become adults, their whole healthcare system changes, on top of now having to do it by themselves – in the cases where guardianship is not needed or obtained. Because of this, he emphasizes the need to teach adolescents in transition how to utilize healthcare, when to call a doctor, when to use the emergency room, how to pick up prescriptions, and more.

At the **Waisman Center University Center for Excellence in Developmental Disabilities (UCEDD)**, YHTI helps make healthcare transition easier for youth with special healthcare needs in Wisconsin by developing and sharing transition resources with families and

“How do I recreate a life for my loved one whether it’s academic, vocational, social, or all of the above?”

-Sheri Edison

A chef in a white uniform and red bandana is cooking in a kitchen. He is stirring a red pot on a gas stove. A tray of meatballs is on the counter next to him.

"You raise kids to become adults. And we want those adults to live a self-determined life." Tim says. "We want them to be able to advocate and stand up for themselves."

FACILITATING ACCESS TO RESOURCES

Sheri came across the Waisman Center website while looking for resources for Blake. She called to learn more about the research being done at the center. “As a parent, you want to be close to the Waisman Center,” Sheri says.

The Edison's and Montoto's experience with transition into adulthood was unique to them, but reflects the difficulties of many families of individuals with IDD to find the right mix of resources and supports. Recognizing the needs of families like the Edisons and the Montotos, the Waisman Center has been focusing efforts on expanding resources, services and trainings to help families navigate the transition to adulthood, making it less siloed, and more cohesive.

Transition to adulthood roadmap developed by WITPP in collaboration with the Rock County community



and the Health Resources and Services Administration (HRSA) grant, has created a program, Transition Counseling Services, through which a group of LEND trainees – in social work, occupational therapy, nursing, and other areas – meet with families to understand their goals, needs, and questions around transition, and find key, curated transition information for them. “It could be things like, ‘I’d like to learn more about what kind of independent living opportunities exist in the community for my child or I want to fully prepare for what to expect financially from adult service systems,’” says **Kate Szidon**, who oversees the program.

The program started around healthcare transition, but has now expanded to include multiple areas. “What we learned is families were interested in learning more about healthcare transition, but they also really wanted to learn about a whole other host of things that were related to adult transition,” Kate says.

Healthcare, education or workforce, social circles and independent living are some of the biggest items on the list of systems or areas of change for individuals with IDD as they become adults. Although these areas can relate to each other, the systems can be quite siloed, and require specific work for each one. In the UCEDD, the Wisconsin Integrated Transition Planning (WITPP) brings together self-advocates, family members, and people who work in education, employment, and healthcare to improve and simplify the transition planning process by promoting integrated transition planning. This refers to thoughtfully mapping out and preparing for the youth’s life as an adult by integrating planning for the youth’s education, work, and healthcare to achieve their goals as an adult.

TRANSITIONING AS A FAMILY

For Lui, it was family support that got him through his battle with cancer. “[Family support] literally helped me out through literally one of my lowest moments of all time. And even when they were down, I felt like I had to get back by just keeping up a positive attitude about the situation as well. So yeah, family support was a very major contributing factor,” Lui says.

For the Edisons, family members, such as cousins, have been key to developing social relationships for Blake, and they hope to rely on them when figuring out independent living situations for Blake. “He’s got six cousins and their spouses who we are bringing into the mix to ensure family support as things move forward. Right now, he lives with us. And we hope that that situation will change in the near future,” Sheri says.

Lui’s hope is to also be able to live independently in the future. “He has ambitions, like any other kid does. He wants his own money. He wants a car,” his dad, Luis, says. “I’ve expressed to him the need for independence,” he adds. He recognizes that as parents, they are not going to be able to be with Lui forever. “I would like for him to be more independent, on his job, his day to day activities, and not depend much on us as parents. Not because it’s cumbersome or it’s bothersome, but because that’s what I guess we as parents always want for our kids, to be independent and to thrive and to be successful.”

“Every year Blake does things that surprise us that you would never think that he would have been able

to do just a year ago or two years ago. So, we continue to keep an open mind and look at Blake just like we look at any person that’s continuing to grow and evolve and learn,” Sheri says. There is a lot of intentional work behind preparing adolescents with IDD for adulthood, in a way that will not just get them through the transition, but provide a sustainable basis for their entire adult life. Although there are many resources to help, they need to be tailored to each person, because there is so much heterogeneity in disabilities, and in life. So, families keep working until they find, as Sheri says, the secret sauce that works for their loved one. ■

CONNECTING FAMILIES: WAISMAN PEER RESOURCES



Connecting Families is a network of peer support in the Waisman Center’s **University Center for Excellence in Developmental Disabilities (UCEDD)** for families of children with special healthcare needs. It nurtures the development of peer support approaches and groups that address unmet needs in the community designed and led by individuals with lived experience in that area.

“Our mission is to hold space and provide culturally, linguistically, and situationally sensitive support and information for families,” says **Gail Chödrön, PhD**, director of the Wisconsin Leadership Education in Neurodevelopmental and Related Disabilities (WI LEND) program and the UCEDD Community Training and Technical Assistance coordinator.

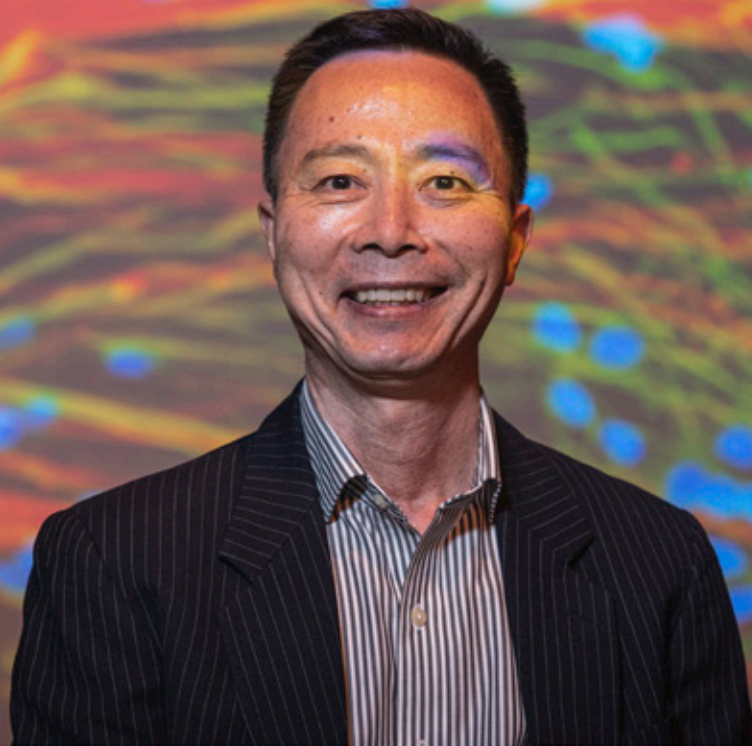
Currently, Connecting Families provides peer support in three areas of unmet need and is developing approaches in a fourth area focused on rural communities, with plans to address additional areas in the future.

The three programs are:

- **Parents Reaching Out (PRO)**
A longstanding peer-to-peer support approach for parents of kids who are deaf or hard-of-hearing
- **Sankofa**
A peer support group targeted specifically to Black families of children with disabilities
- **Children with Complex Medical Needs**

For more info and to connect go to
connectingfamilies.waisman.wisc.edu/

Transitions



After more than two decades at the Waisman Center, **Su-Chun Zhang, MD, PhD**, professor of neuroscience and neurology, retired. Zhang changed the landscape of stem cell research with several first-in-the-world scientific breakthroughs including being the first to coax stem cells into becoming neurons and astrocytes. His work on a potential treatment for Parkinson's disease is currently under review for a clinical trial. Zhang plans to continue his research with BrainXell, a private company he founded.

photo by Andy Manis



David Wargowski, MD, clinical geneticist, retired after 34 years at the Waisman Center Clinics. "I always wanted to help people and knowing that I have been able to, is very humbling and rewarding," Wargowski says. "You always hope that you can have an impact on someone's life and I am deeply thankful for the ability to have done so."



Steve Marshall, MS, MSPH, is the new associate director for finance and administration. Marshall, previously the division administrator of infectious diseases at the UW School of Medicine and Public Health (SMPH), brings extensive expertise in grant writing and research administration. The position was previously held by **Jody Bleck, MS**, who retired.



The **Waisman Early Childhood Program (WECP)** welcomed **Teresa Hoveland** as its new director in September. Hoveland brings more than 20 years of experience in campus childcare, early childhood development, and programmatic leadership. Hoveland student taught in the WECP while an undergrad at UW-Madison.

Q&A



*Lily Wagner, PhD, BCBA, is a licensed clinical psychologist and the new director of the **Waisman Center Autism and Developmental Disabilities Clinic**. Wagner joins the Waisman Center from Vanderbilt University Medical Center in Nashville, Tennessee, where she was a part of the Treatment and Research Institute for Autism Spectrum Disorders.*

TELL US ABOUT YOURSELF AND YOUR JOB.

I specialize in autism spectrum disorders where I do diagnostic work. We see a range of ages in the A&DD Clinic. I specialize in working with little kids usually 18 months to three-years-old suspected of having autism or other developmental delays or concerns. I also do research. Previously, my research has been related to identifying new tools and systems for early identification, especially in communities that traditionally face disparities such as rural families, non-English speaking families, and Black families.

WHAT WAS YOUR PATH TO CLINICAL PSYCHOLOGY?

I just kind of fell into it. I took a really circuitous path. I was a professional ballet dancer. And then I worked in advertising for a while in New York City. And then I just wasn't really feeling fulfilled in that role. So, I went back to school and I got my master's in special education at Vanderbilt and became a board-certified behavior analyst (BCBA). But then as I was finishing up that master's program, I decided to pursue a doctorate in psychology at the University of Georgia. My doctorate is in school psychology, actually but all of my practicum and internship experiences were clinical. And through those experiences, I really became interested in autism. That interest led me to pursuing my postdoc at Vanderbilt, which was at the Treatment and Research Institute for Autism Spectrum Disorder where I became really involved in research.

WHY DID YOU FOCUS ON AUTISM?

The presentations look so different across children and across genders too. There's so much that we're learning about autism with the research being updated so frequently. It's easy to stay interested when it's not a stagnant field. Then the prevalence continues to increase but we're not keeping pace with being able to offer both diagnostic and treatment services. So, we have this huge gap in care, which I'm really motivated to address and find new ways to try to help. I just think a lot of our old systems and tools that we've used for a long time are just not appropriate necessarily for every kid anymore. And we need to be thinking of new solutions to try to help these families faster.

WHAT BROUGHT YOU TO THE WAISMAN CENTER?

Waisman obviously has a great reputation and I was looking for a leadership position. I had also heard so much about Leann DaWalt, PhD, who is the director of the University Center for Excellence in Developmental Disabilities (UCEDD). She's worked with a couple colleagues of mine and I knew that she would be a great mentor for me in terms of helping foster my research and helping connect me to community partners. Just being able to not only do clinical work but also get leadership experience and continue my teaching and research, has been great. When I first came here I also loved everyone that I met. They were all so friendly and welcoming which is very important.

WHAT DO YOU LOOK FORWARD TO AT WAISMAN?

I'm really looking forward to building relationships with community partners that we don't have right now. Then I did a lot of work with our statewide Birth to Three program in Tennessee where we had a lot of research collaborations with them and that was a great way to help push out some resources to families. I'm hopeful to start building a deeper relationship with our Birth to Three programs here in Wisconsin. I'm really excited to explore tele-assessment in ways that we might be able to integrate it within our A&DD Clinic. I'm also excited to do some quality improvement initiatives around our existing visit types and understand what's working well and how we could maybe expand those offerings or modify them to work best for our community.

WHAT DO YOU LIKE TO DO IN YOUR SPARE TIME?

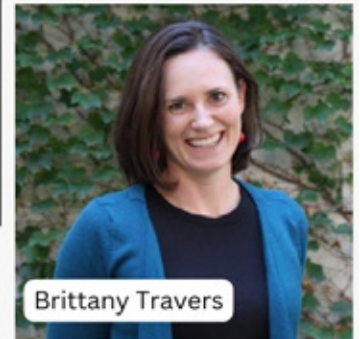
I have two young daughters, so they take up a lot of my time, of course. I am also an avid runner. I love to run which has been great because Madison happens to have so many great trails and things like that. My family and I love to go hiking too, so that's really fun. We also love to try new restaurants, so I've been asking everyone for their restaurant recommendations. We're just excited to be outside and explore as much as possible.

Where in the World are Waisman Fulbright Scholars?

Being awarded a Fulbright is a globally recognized and highly prestigious achievement as it is one of the top awards in all of academia. The program's purpose is to expand perspectives through academic and professional advancement and cross-cultural dialogue by sending accomplished graduating college seniors, graduate students, and young professionals to more than 140 partner countries to conduct research or teach English.

2022

Brittany Travers, PhD, Waisman investigator and associate professor of kinesiology spent six months in Malaga, Spain at the University of Malaga where she taught a graduate class on neuroplasticity and collaborated with researchers on various projects.



Brittany Travers

Spain

2024

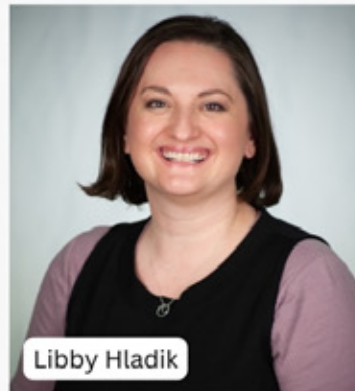
Ben Parrell, PhD, Waisman investigator and associate professor of communication sciences and disorders, will continue his research in Spain where he will be looking at the impact of ataxia – a neurodegenerative disease – on speech in Spanish.



Ben Parrell

2024

Libby Hladik, PhD, former Waisman graduate student, left for the Czech Republic where she is currently conducting research at Masaryk University in Brno. She is working on developing culturally-appropriate assessments for disability inclusion in cultural institutions such as museums and libraries.



Libby Hladik

Czech Republic

Australia

2022

Brittany St. John, PhD, MPH, OTR/L, former Waisman graduate student and now assistant professor at the University of Washington, spent over half a year at Latrobe University in Melbourne, Australia. She piloted a six-month caregiver-mediated feeding intervention for kids with autism.



Brittany St. John

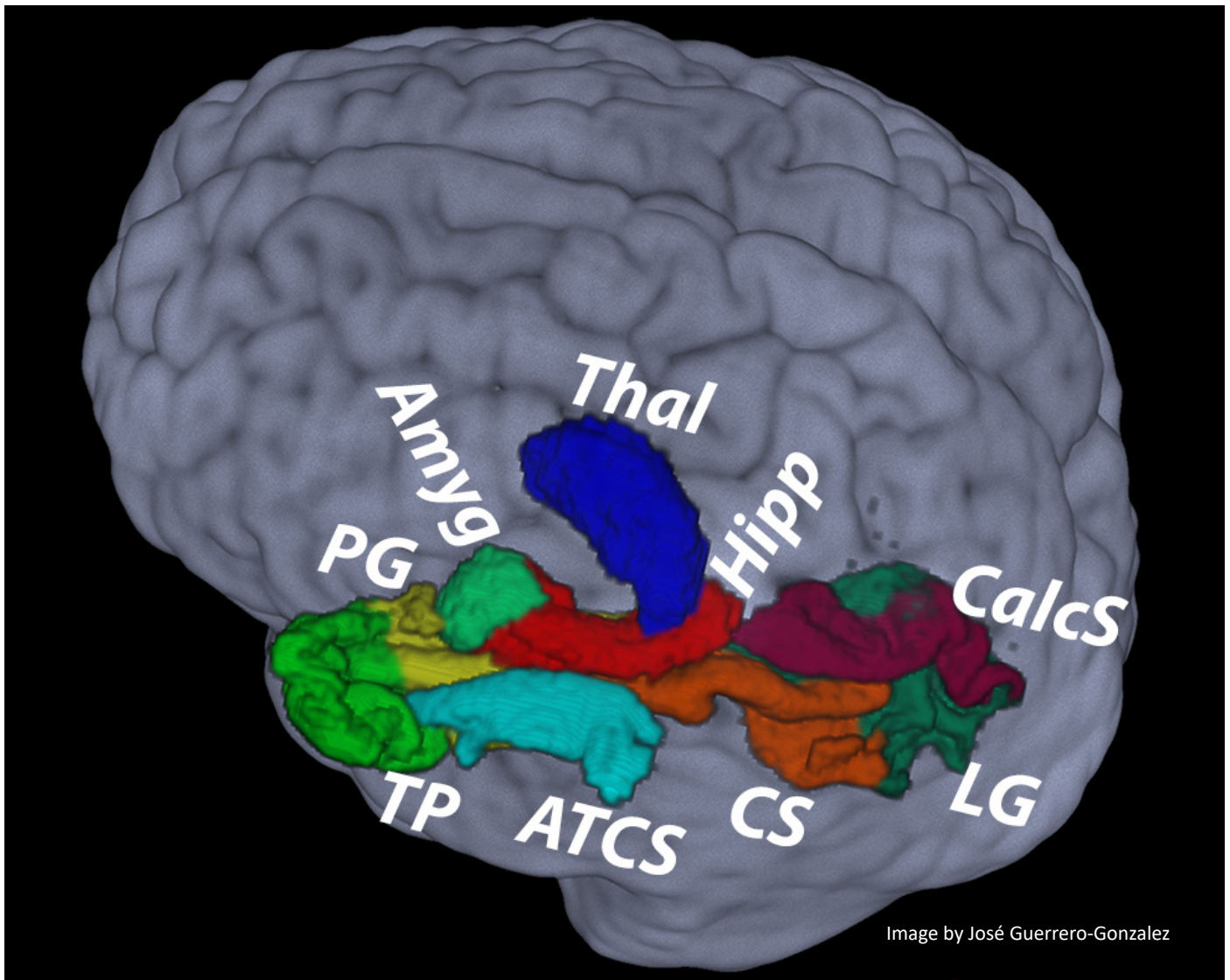


Image by José Guerrero-Gonzalez

Memory Impairment After Severe Traumatic Brain Injury in Adolescents Related to Size and Connectivity Changes in the Hippocampus and Beyond

By Charlene N. Rivera-Bonet

Memory impairment as a result of a traumatic brain injury (TBI) has long been associated solely with the hippocampus, the brain's learning and memory center. However, new research published in the journal *Brain Imaging and Behavior*, reveals that the hippocampus is not acting alone. The study from the University of Wisconsin-Madison's Waisman Center, indicates a broader network of brain structures is involved in TBI-induced memory impairment in adolescents.

Traumatic brain injuries usually result from a violent bump, blow or jolt to the head or body, or an object going through brain tissue. The hippocampus, found deep in the human brain, is highly vulnerable to TBI regardless of the main site of the initial injury.

The study, led by **Peter Ferrazzano, MD**, Enid and Jerry Weygandt Professor of Pediatrics and director of the Pediatric Brain Care Clinic at the Waisman Center, found that although the hippocampus has been in the spotlight for leading to impaired memory after a TBI, memory performance in children relates to the integrity of the entire memory network,

(Image at left) Brain regions with structural connection to the left hippocampus (Hipp). Abbreviations: parahippocampal gyrus (PG), thalamus (Thal), collateral sulcus, (CS), amygdala (Amyg), temporal pole (TP), calcarine sulcus (CalCS), anterior transverse collateral sulcus (ATCS), lingual gyrus (LG).

not just the hippocampus. This is part of a bigger effort to identify markers that can predict outcomes in children with severe TBI. “We designed a prospective study of these children [who experienced a severe TBI] one to two years after their brain injury events to try and understand how the changes in their brain are related to their outcomes,” says **Andrew Alexander, PhD**, professor of medical physics and psychiatry, and one of the authors of the study. “In this study, we were focused on how the hippocampus and networks associated with the hippocampus are affected, and how that relates to working memory in these kids.”

The researchers did a comprehensive study of the memory network in relation to memory and learning performance in adolescents with a severe TBI, one to two years after their injury. They measured memory and learning performance, and used magnetic resonance imaging (MRI) to look at brain structure and function. In addition to evaluating multiple parts of the memory network – the fornix, thalamus, and the calcarine sulcus – they used multiple measures such as size, structural connectivity and functional connectivity, making it a comprehensive assessment of the brain after injury.

Their results showed the hippocampus was smaller in size in adolescents with traumatic brain injury compared to individuals without injury. This loss of hippocampal volume was consistent with worse performance on memory and learning in individuals with TBI.

Structural connectivity measures look at direct, physical connections between brain regions. It assesses the state or condition of the fibers that act as a bridge to connect the hippocampus to other parts of the brain. Adolescents with TBI, the study shows, have structural abnormalities in prominent fiber bundles that connect the hippocampus to other brain regions, including the fornix. These abnormalities were related to worse performance in the memory and learning tests.

As opposed to structural connectivity, functional connectivity measures the synchrony, or lack thereof, between regions of the brain that do not necessarily have

a direct physical connection. Even without a straight, physical connection, regions can share signals through indirect ways, and their ability to signal each other is important for brain function. Results from this study show that the hippocampus was less structurally and functionally connected to other important regions of the memory network – the thalamus and calcarine sulcus, with effects on worse performance of memory and learning.

The thalamus and the calcarine sulcus are involved in sensory processing and vision, respectively. These regions have strong connections to the hippocampus and are involved in guided attention, and may be important for “visualizing” a word when trying to recall it. “That was a very interesting finding. Because when these participants

are asked to remember the words on the test, they may be forming mental pictures of this task, or objects that they're asked about,”

explains **José Guerrero-Gonzalez, PhD**, lead author of the study.

The population with traumatic brain injuries is a heterogeneous one. “There's really no two cases that are identical,” Alexander says. “So, by taking a more holistic approach to looking at multiple measures we can develop a more comprehensive picture of what's going on with the hippocampus as well as the network associated with the hippocampus.”

“Those were the main takeaways, that not only the hippocampus, but the network would be altered by the injuries, so there would be effects observed on the whole network,” Guerrero-Gonzalez summarizes.

These effects being measured one to two years post injury means that the association between hippocampal atrophy and memory dysfunction continue in adolescents with severe TBI years after the injury. However, following injury, the hippocampus may have some capacity for reorganization and potential for the formation of new neurons. Broadening research to focus on areas beyond the hippocampus may point to other potential mechanisms that aid or impair recovery after a TBI. ■

“... not only the hippocampus, but the network would be altered by the injuries.”

-José Guerrero-Gonzalez

When Worlds Collide: Autism & ADHD

Working to better understand, diagnose,
and treat co-occurring autism and ADHD

By Emily Leclerc

Note: Both the Li Lab and Travers Lab use identity-first language in response to the growing preference for that language in the autism community. Both Felix and Erin prefer person with autism, so person-first language will be used when referencing Felix.

Felix Benton is a bright and bubbly eight-year-old with more energy than he or anyone else knows what to do with. “Fe (Felix’s nickname) likes to say that he has energy boxes up to Mars,” Felix’s mom, Erin McConnell, says. Constantly bouncing around and looking for something to get into, Felix is a ball of love, curiosity, and enthusiasm. He is currently just a little bit obsessed with engineering and science and spends a lot of time asking questions about how things work.

“He always has these deep questions right before bedtime. They can be about the day or about something ‘sciencey’ he has learned or even just what if things were this other way,” Erin says. Felix is a smart and vivacious kid with no fear of being his true self. “He is so himself like in how he dresses, how he acts, how he speaks, how he finds humor in things. He is just such a gift,” Erin says.

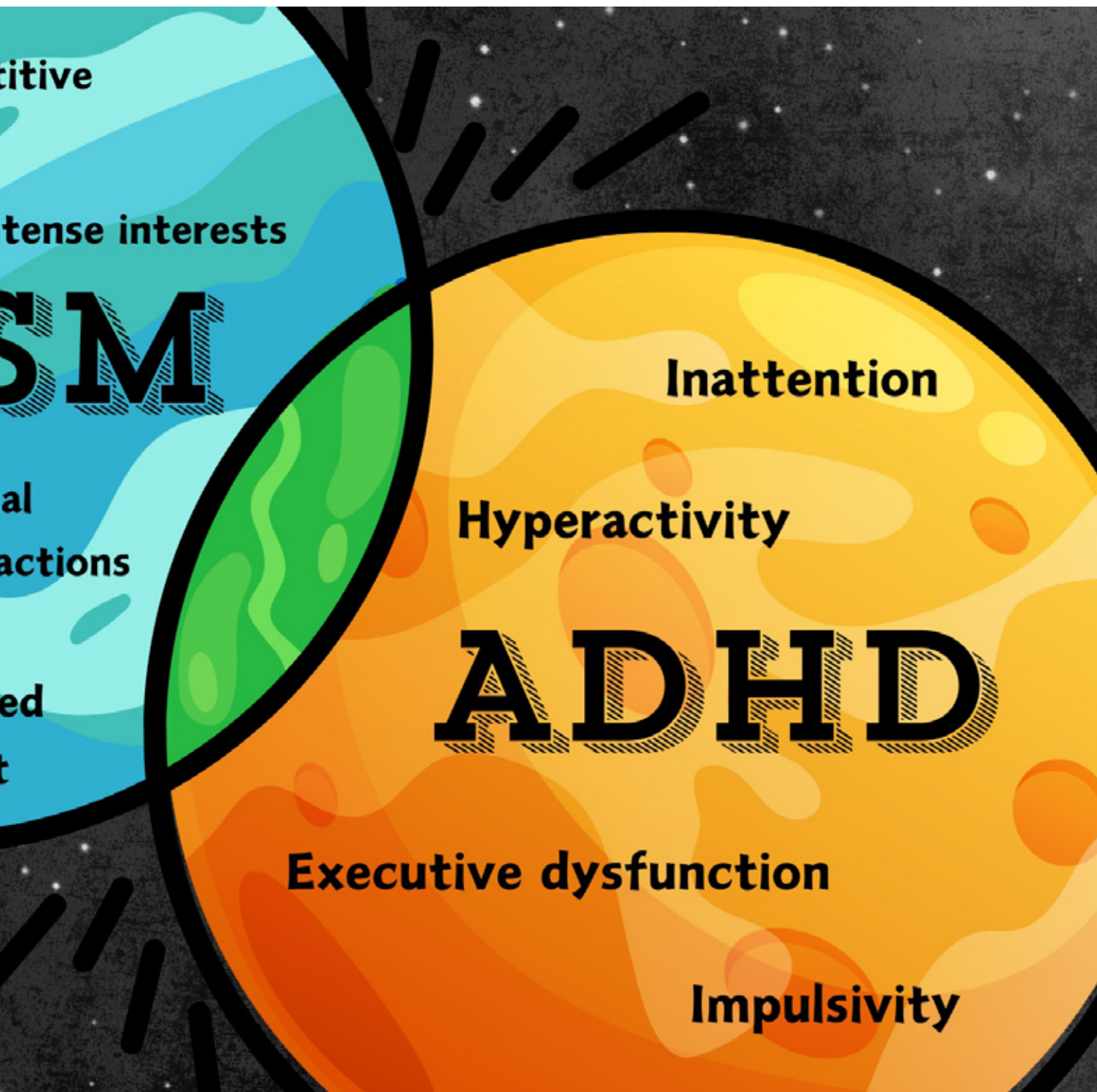
Felix bounces through life with an eagerness all his own. He also has to work through some challenges that not all kids face. When Felix was younger, he was diagnosed with autism and ADHD. This combination can present unique challenges in day-to-day life for Felix.

Autism symptoms can vary greatly in both severity and presentation as they exist across a spectrum. According to the



Centers for Disease Control and Prevention (CDC), “Autism spectrum disorder (ASD) is a developmental disability caused by differences in the brain. People with ASD often have [issues] with social communication and interaction, and restricted or repetitive behaviors or interests.” Similar to autism, ADHD is also a neurodevelopmental disorder that originates during childhood and is characterized by developmentally inappropriate levels of either inattention or hyperactivity and impulsivity, says **James Li, PhD**, Waisman investigator and A. A. Alexander Associate Professor of Psychology and Psychiatry.

Autism and ADHD actually co-occur rather frequently. It is estimated that 40% of children diagnosed with autism



will also be diagnosed with ADHD. On their own, autism and ADHD can heavily impact a person's day-to-day life. But diagnosed together, the two can exacerbate the other's symptoms, adding even more barriers and challenges than a single diagnosis.

"Having ADHD alone can be impairing. One of the hallmarks of having it is that the individual has a hard time with either being attentive to things they need to be attentive to or they are hyperactive and can't sit still. That can cause school-based difficulties, peer relationship difficulties, and even familial problems," Li says. "For an autistic child, they might already have a harder time in certain situations or certain social contexts. Then you add ADHD symptoms on

top of it – like the inattention and hyperactivity – and that just exacerbates their abilities to regulate and benefit from social interactions with others."

And on top of all of that, co-occurring autism and ADHD is poorly understood and very understudied. Autism and ADHD could not even be diagnosed together until 2013. Up until then, the diagnoses were considered mutually exclusive – if you were diagnosed with one then you could not be diagnosed with the other. This made it virtually impossible to receive funding to study the two together. When the guidelines finally changed in 2013, the research was far behind and has spent the last decade trying to catch up.



Felix Benton with one of his chickens.

There are also no standard treatments for the combination of autism and ADHD and many treatments currently available are less effective when both conditions are present. “We’ve seen, based on some data collected over the last several years, that when you give an ADHD treatment to an autistic child it is not going to have anywhere near the same efficacy as giving that treatment to a child without autism,” Li says. The psychosocial interventions used to treat autism also seem to be greatly affected by the child also having ADHD. Currently, there are no evidence-based psychotropic or psychosocial interventions and treatments for kids with autism and ADHD.

This makes it even more difficult for clinicians, school teams, and family members to figure out the best ways to support kids with both diagnoses. It has taken Erin, and the rest of Felix’s family and support team, many years to help him build and implement strategies to make it easier for him to navigate the world with many ups and downs throughout his life.

This lack of information on autism and ADHD co-occurrence not only leads to kids facing more challenges and struggles but also ineffective treatments and therapies. That understanding was the impetus behind why Li and **Brittany Travers, PhD**, Waisman investigator and associate professor of kinesiology, decided to launch the University of

Wisconsin Longitudinal Imaging and Neurogenetics in Kids Study or UW Link Study.

The study’s goal is to investigate the biological underpinnings of co-occurring autism and ADHD in order to understand why the two occur together so frequently. “It seems like there has to be some type of biological aspect to this that we can hope to better understand through this project. Then we can better conceptualize what is co-occurring autism and ADHD and also hopefully have targets to be able to think about whether there are ways we can ameliorate some of those features,” Travers says. Li and Travers are working to lay the foundation for future work into developing more effective treatments, therapies, and interventions.

Felix is a participant in the UW Link Study and Erin says he looks forward to every visit. He is absolutely thrilled to be involved with science and gets even more excited when the researchers ask him questions about himself. Erin knows that the study’s results, once it concludes, may not help Felix right now but it will be able to help all of the future kids who get diagnosed with autism and ADHD.

“I think about how difficult a lot of Felix’s experiences have been and will continue to be. But he is making the choice to be part of a study that helps us understand what is going on, provide more resources for the future, and develop

language for how the neurodivergent experience interfaces with the world,” Erin says. “As the discourse gets more open, and everyone at Waisman continues their amazing work, if we can kind of pay it forward and push those benefits into the future, we would all be really excited and grateful for that.” The Waisman Center isn’t the only place researching ADHD and autism either. Work is occurring around the globe to further our understanding of the two conditions and how best to provide the support and resources that the kids need.

Alongside this boom in research, the diagnostic process for autism, ADHD, and related mental health conditions is seeing change as well. Typical diagnoses are done by following the guidelines set out in the DSM (the Diagnostic and Statistical Manual of Mental Disorders), which is the handbook used by healthcare professionals throughout the U.S. The DSM outlines the symptoms and criteria for an official diagnosis. The criteria are very straightforward and do not consider the variances and nuances of mental health. This system can be challenging for individuals with conditions that are on a spectrum – such as autism and ADHD – where individuals very often may not fit neatly into diagnostic boxes.

“We can’t just put people in arbitrary categories based on do you have six or more symptoms of this condition. If it’s six, great you have it. Oh, but you only have five? That means you don’t have it. It’s arbitrary and doesn’t make sense biologically,” Li says. “The person with five symptoms may still be impaired and struggling and the way we define it makes all the difference in the world. It’s the difference between whether that person with five symptoms gets supports and resources or doesn’t.”



James Li

To address the DSM’s limitations, a consortium of researchers proposed a new diagnostic method – HiTOP. HiTOP stands for the Hierarchical Taxonomy of Psychopathology and focuses on dimensional views of conditions rather than categorical.

“The fundamental goal of HiTOP is to shift the narrative that mental disorders are discrete categories that people fit neatly into. Instead, we want to think about where individuals function across different dimensions of mental health. It’s all about the gray areas and where people fall on them,” Li says.

HiTOP would allow clinicians to look beyond a black and white label to the full gamut of symptoms to see the individual’s challenges and successes. Then treatments and therapies can be targeted to a person’s greatest needs rather than a generalized approach. HiTOP better represents the large variation seen in mental health and human diversity.

While not currently used in clinical settings, Li is one of many researchers talking and educating about HiTOP to hopefully make it a diagnostic option in addition to the DSM. It could improve clinicians’ ability to help kids with autism and ADHD because they can more specifically determine where the child may need help and where they don’t. It would also help loosen the constraints seen in DSM diagnoses and potentially aid more kids in getting the necessary supports.

Today, Felix still has struggles in school and at home even with a robust IEP (individualized education plan) in place. His co-diagnoses present him with challenges and they likely will for the rest of his life. But none of that stops him from living life to its fullest, being an absolute bundle of joy, and a delight to be around.

While having the diagnoses are helpful for navigating the system, having language to more accurately discuss his experiences, and getting him the access to the supports that he needs, Erin emphasizes that they don’t place a lot of stock in those labels. They are just words after all. Felix is so much more than his diagnoses and he should be recognized as such. “People who have ADHD and autism are often incredibly creative and empathetic. They can make really unique connections, provide different insights, and make amazing friends. They can have quite the sense of humor too. And sometimes people forget that these are qualities that neurodiverse people can have,” Erin says. “Felix embodies all of that and is just so himself and that is such a great gift.”

So, while the research works on developing a better understanding of co-occurring autism and ADHD and HiTOP works to improve the diagnostic process, Felix will continue to throw himself off playground equipment, ask a million questions, wonder why things work the way they do, and be a bright effervescent kid. Erin and the rest of Felix’s family will follow his lead, doing everything they can to help him have the best life possible. ■

Taking Out the Trash

Study finds clearing specific cell “trash” is possible and may be target for future treatments of neurodegenerative diseases

By Emily Leclerc



Luigi Puglielli
photo by Andy Manis

Cells make a lot of trash. Probably more than you’d think. So, cells have a trash disposal system that efficiently cleans up and recycles any waste that is produced. But when there are issues with the disposal system and trash builds up in the cell, that can lead to disease. This has led researchers to consider the trash disposal system as a target for potential disease treatments and therapies. New research from the Waisman Center reveals that stimulating a cell’s disposal system in specific places could potentially be an effective treatment route for diseases that are caused by trash accumulation, specifically misfolded proteins.

Waisman investigators **Luigi Puglielli, MD, PhD**, professor of medicine, and **John Svaren, PhD**, Vilas Distinguished Achievement Professor of Comparative Biosciences, recently published a study that showcases a new way to help clear away misfolded protein buildups specifically in a cell’s endoplasmic reticulum that are associated with Charcot-Marie-Tooth disease (CMT) by stimulating a cell’s trash disposal system. The paper, Spatial selectivity of ATase inhibition in mouse models of CMT disease, was published in the journal *Brain Communications*.

The endoplasmic reticulum (ER) is a structure found inside every cell in the body that is responsible for producing many of the proteins a cell needs to function. As part of the production process, proteins will fold into their final shape inside of the ER. To proteins, shape is everything. Only a properly folded protein can function correctly. A protein that folds into the wrong shape has to be diverted to the trash disposal system to be broken down

and recycled. Misfolded proteins are tagged for the trash by the ER’s quality control system – the acetylation machinery being an important part of it.

The ER acetylation machinery, which was discovered and defined by Puglielli and his lab, ensures that proteins inside the ER are folded correctly and if they aren’t, it activates autophagy. Autophagy is one part of a cell’s trash disposal system. It is the process through which the cell breaks down waste and recycles it.

When there is a mutation in the genome that causes proteins being made inside the ER to misfold, they often collect together into clumps called aggregates. Autophagy has a tough time clearing away protein aggregates for multiple reasons. A new theory for potential treatments to conditions resulting from aggregates such as Charcot-Marie-Tooth disease (CMT), Alzheimer’s disease, Amyotrophic Lateral Sclerosis (ALS), and Huntington’s disease, is to stimulate autophagy to help degrade and recycle the protein aggregates. The trick is finding a way to activate autophagy in the correct place.

“There is a lot of interest in using tools like biochemical compounds that stimulate autophagy to resolve disease because it can help cells to digest protein aggregates. Like helping to clean up the trash in the house,” Puglielli says. “If you have protein aggregates in the ER or another organelle we want to be able to activate autophagy and limit the activation to a specific place. For example, if you have trash in the kitchen, you don’t start cleaning the bathroom. You go right to the kitchen.” Generalized activation of autophagy can have a number of side effects that can be damaging to the cells and to the individual as

a whole. Determining ways to activate autophagy exactly where it is needed will allow researchers to utilize its waste cleaning abilities while avoiding harmful side effects.

Puglielli focused on two proteins – acetyltransferase 1 (ATase1) and acetyltransferase 2 (ATase2)— that are a part of the acetylation machinery in the ER to see if they can be used to selectively stimulate autophagy inside the ER and nowhere else. ATase1 and ATase2 regulate the activation of autophagy inside the ER. By blocking the ATases, Puglielli was able to activate autophagy inside the ER. To determine if the ATases only stimulate autophagy inside the ER, Puglielli and Svaren, whose research focuses specifically on CMT, set up two different mouse models of CMT – one where the protein aggregates form outside the ER and another where they form within.

Charcot Marie Tooth disease, or CMT, is an inherited nerve disorder that impacts nerves in a person's feet, legs, hands, and arms. There are several different types of CMT that are categorized by what type of genetic mutation an individual has. According to Svaren, roughly 10% to 15% of CMT cases are associated with protein misfolding caused by different mutations.

Puglielli found that when they stimulated autophagy by blocking the ATases, autophagy only activated inside the ER. In the mouse model where the protein aggregates formed outside of the ER, the aggregates were unaffected. “This system can discriminate between events inside the ER and outside the ER,” Puglielli says. This discovery establishes the ATases as the first target for addressing protein aggregates that form inside of the ER. It activates the cell's trash disposal system and in one selective space.

This work provides important foundational information for potential personalized CMT therapies down the line

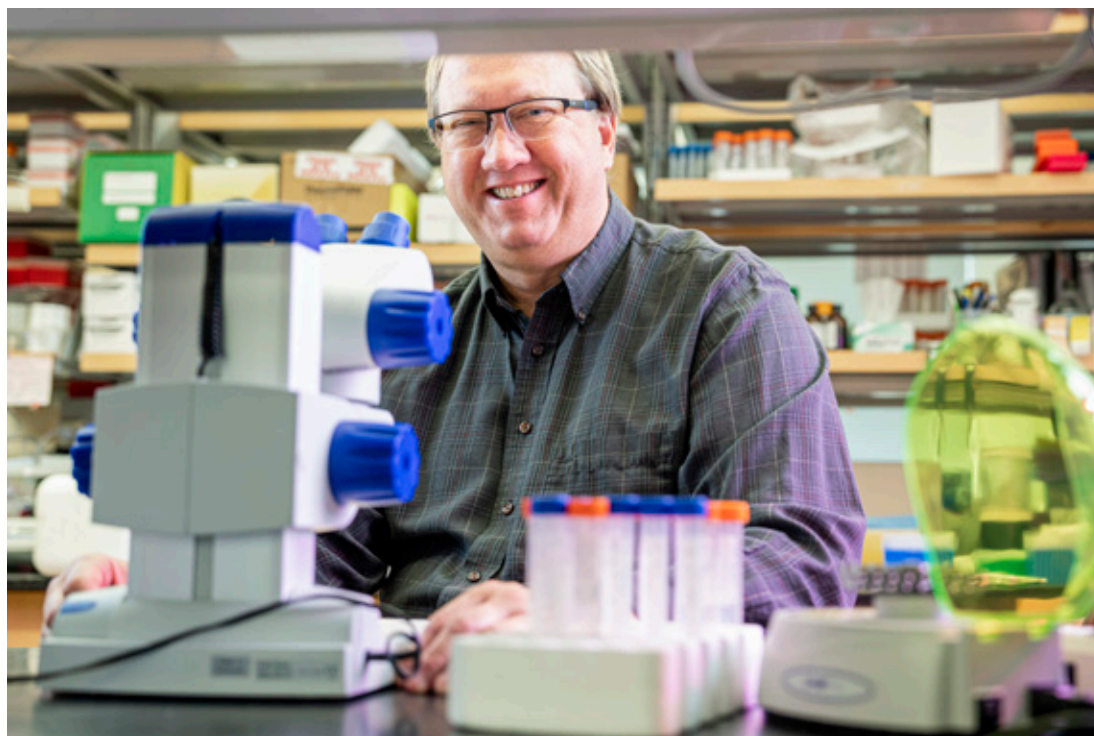
– and potential therapeutic avenues for other conditions caused by protein aggregates. Cystic fibrosis – a genetic condition that causes often severe damage to the lungs, digestive system, and other organs – is also caused by protein aggregates inside of the ER. Puglielli's discovery may provide another option for a potential treatment for this chronic condition. It also expands knowledge of selective autophagy activation that can be applied to a broader range of conditions where aggregates form in other places outside of the ER.

“The NIH (National Institutes of Health) said recently that there are 15,000 rare disorders. And a lot of dedicated foundations are focused on trying to find individual treatments for all of these disorders. But what if we can find common attack points for multiple rare disorders,” Svaren says. “Sometimes there are common mechanisms that occur in different cell types and organs that cause different diseases but are all based in one fundamental cause. If it's something fundamental, like autophagy, you could potentially attack several diseases at the same time.”

While this work is far from being translated into actual therapies, the research doors it opens are important. From here, Puglielli wants to build an effective pipeline to work on finding a compound or drug suitable for clinical trials that accomplishes this selective autophagy through ATase blocking. That could take some time but he is excited by the prospect of translational research. Svaren looks forward to the opportunity to again work alongside Puglielli in pursuit of improved therapeutics for CMT and more.

“Puglielli really discovered this whole area himself and has developed the first pharmacology [research compounds] in this area as well,” Svaren says. “It has promise in a variety of diseases and it looks like it has real promise for CMT as well. That is exciting.”

This research was funded in part by the Charcot-Marie-Tooth Association – a nonprofit organization determined to bring researchers, clinicians, patients, and companies together to help accelerate the development of treatments and hopefully find a cure for CMT. ■



John Svaren

photo by Andy Manis



Improved Assessment Protocols Identify Cerebral Palsy Earlier

By Emily Leclerc

Research from the **Pediatric Neuromodulation Lab of Bernadette Gillick, PhD, MSPT, PT**, at the Waisman Center showcases that evidence-based assessments for cerebral palsy (CP) are helping to improve diagnostic timelines and access to therapies and interventions. These assessment pathways, recommended by clinical guidelines published in 2017, were intended to help facilitate earlier CP diagnoses. Gillick, a Waisman investigator and professor of pediatrics, investigated the implementation of new assessment protocols implemented in 2017 to evaluate their impact on how early an at-risk child was diagnosed with cerebral palsy and how that affected the family's access to interventions. "The importance of developing pathways to identify children at high-risk for CP cannot be understated, early detection leads to early intervention and the promise of improved lifelong outcomes," says Gillick.

CP is the leading cause of childhood disabilities in the United States. It is a varied neurological condition that affects body movement and muscle coordination. CP is a secondary diagnosis associated with either a brain injury or other atypical brain development in early infancy. Early identification and diagnosis of the condition is important as it allows for earlier access to resources and supports such as treatments, therapies, equipment, and financial support.

"When physicians are able to provide an early CP diagnosis, this may help give families an understanding of what supports might be needed for their child, get access to more cerebral palsy-specific information, and understand how their child may benefit from certain therapies and early interventions," says **Ellen Sutter, PT, DPT, PhD**, lead author on the related recent study publication and former graduate student in Gillick's lab. She is now a postdoctoral researcher at Boston University. Early diagnosis has also been associated with better long-term health outcomes in individuals with CP.

The paper, Evidence-Based Infant Assessment for Cerebral Palsy: Diagnosis Timelines and Intervention Access in a Newborn Follow-up Setting, which was

published in the *Journal of Child Neurology*, reviews data from the **Waisman Center's Newborn Follow Up Clinic** from 2010 to 2022 to determine if the implementation of the new assessment protocols starting in 2017 improved diagnostic timelines and access to interventions. Key assessments included magnetic resonance imaging (MRI), the General Movements Assessment which evaluates an infant's spontaneous movements, and the Hammersmith Infant Neurological Examination, which is a standardized neurologic assessment.

"In general, when children received more of these evidence-based assessments, they tended to receive earlier diagnoses and families were told about the likelihood of cerebral palsy earlier. This paper provides additional evidence that these assessments are helping to support providers in making earlier diagnoses," Sutter says.

This study helps to add more evidence that this series of assessments for CP are effective and worthwhile. This then, in turn, encourages clinics and clinicians to access the training required to appropriately deliver the assessments.

The assessments included in the paper can have costly training from both a time and resource perspective. But with the understanding that they are capable of providing accurate and earlier CP diagnoses, it shows clinicians that the training is worth the investment in time and resources as the assessments can directly benefit patients and improve long-term outcomes.

There is still work to be done but Gillick and Sutter hope that this work will help pave the way for improved diagnostic journeys of individuals with CP and streamlined access to interventions and resources. ■



“The future is not some place we are going to, but one we are creating. The paths to it are not found, but made; and the activity of making them changes both the maker and the destination.”

— Peter Ellyard

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