



The chatter  
within our  
cells  
*Page 2*

Waisman  
Center  
In the News  
*Page 3*

Upcoming  
events  
*Page 4*

A SOFT  
Conference  
*Page 5*



## Helping children live healthier, happier lives

**Sam Thompson** is two-and-a-half years old and a super-charged giver of hugs.

His green eyes sparkle as he first embraces his physician, **Jessica Scott Schwoerer**, and then his dietitian, **Nikki Drilias**. Then it's a quick, laughter-filled sprint across the room to his parents, Josh and Angie, who also get huge hugs.

Sam is at the **Waisman Center Biochemical Genetics Clinic**—a partnership with UW Health and the American Family Children's Hospital—for a scheduled checkup. It's part of his treatment for a metabolic disorder called very long-chain acyl-CoA dehydrogenase deficiency, or VLCAD.

Individuals with VLCAD are unable to break down some kinds of fats, especially during gaps between meals. Sam's VLCAD was picked up by Wisconsin's Newborn Screening Program, which tests for 46 disorders, including metabolic conditions.

The majority of metabolic disorders treated at the clinic, such as VLCAD, have no cure. Instead, "they require lifelong nutritional management," says Drilias, "and our first goal, when we meet families, is educating parents about how to manage and monitor their children's diets." Sam, for instance, has to be on a low-fat diet and drink supplements with milk three times a day.

At the clinic, parents meet and work with a multidisciplinary team of physicians, nurses, genetic counselors and dietitians. They learn about the disorder their child has been diagnosed with and how to manage the treatment on a day-to-day basis. It can be an overwhelming experience.

"The first year was a tough one, with getting Sam to eat enough and the short times between meals"

says Josh. "We would call Nikki (Drilias) all the time, and she was always available to help us."

As children grow older, clinicians spend more time educating them so that they can become medically independent and manage their special diets themselves.

"It's great to watch these kids grow up – children who may have not survived or developed typically in the past are now thriving," says Scott Schwoerer.

Maintaining these lifelong relationships helps patients as well. Josh and Angie live near Milwaukee, and it's about an hour's drive each way to the clinic from their home. "We just don't want to leave this team," says Angie.

Clinic team members also participate in outreach clinics throughout Wisconsin to provide invaluable genetic services that may not be available in all areas. They also host patient and family events to build and support networks among those who may be impacted by the same genetic conditions.

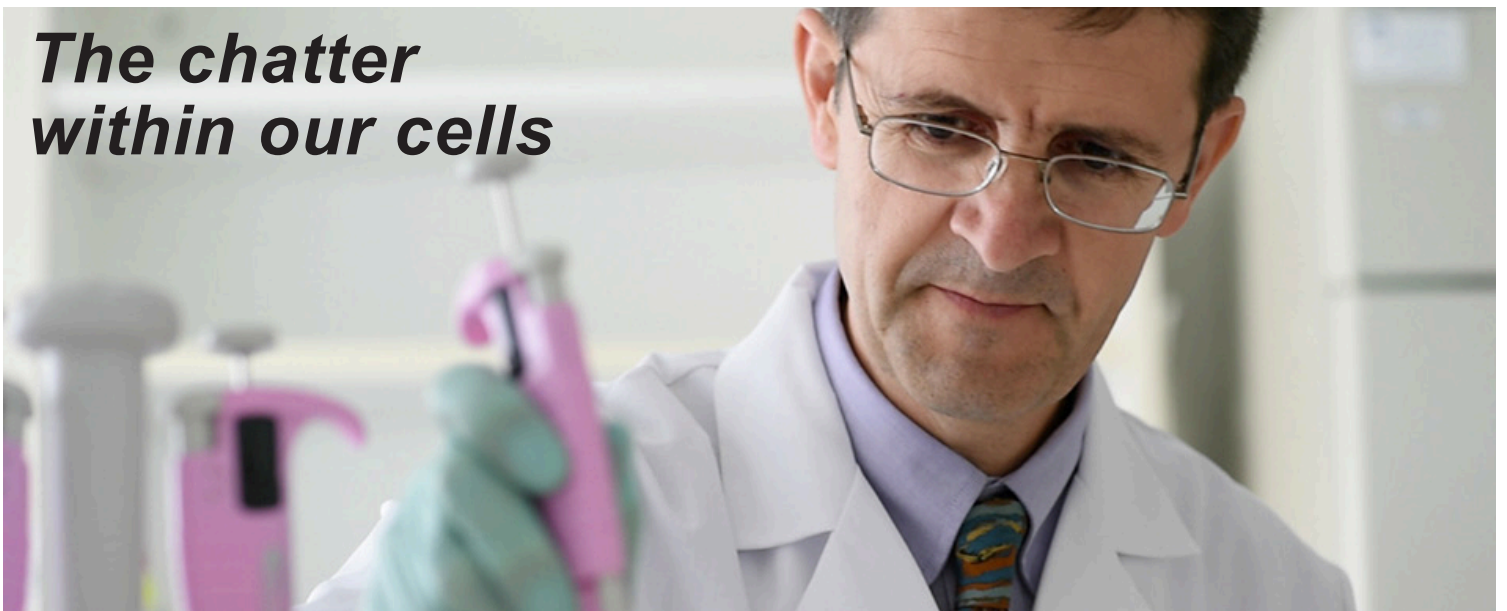
The Waisman Center also has a rich legacy of research and training on metabolic disorders. Clinic team members further this legacy by training medical students, residents and fellows, as well as genetic counseling students and trainees.

Ultimately, it's all about the patients, says Scott Schwoerer. "For all of us at the clinic, what pushes us to put in our best efforts is knowing that we are helping keep children happier and healthier than they otherwise would have been."

The hugs she gets from Sam are an added bonus.

To read the unabridged story, please visit [waisman.wisc.edu/newsletter.htm](http://waisman.wisc.edu/newsletter.htm)

# The chatter within our cells



**Waisman Center** investigator **Luigi Puglielli** had trouble with only one class in medical school: biochemistry.

Yet today he uses biochemical approaches to study some of the vital molecular mechanisms that regulate brain physiology and pathology across the lifespan.

Puglielli, who is a professor in the Department of Medicine, focuses on trying to understand how proteins are tagged with a chemical label through a process called acetylation in specific areas within our cells.

Acetylation of proteins has many functions. For example, it is “important for the development of the brain but also plays a role in several neurodegenerative diseases that affect the brain,” says Puglielli. “Mutations that affect acetylation turn out to be significant factors for various disorders, including autism spectrum disorder and intellectual disability, spastic paraplegia, and Alzheimer’s disease.”

Scientists believed that proteins were acetylated in two locations within cells: the nucleus, which contains our DNA and serves as the ‘control center’ of the cell, and the cytosol, which is the soupy environment surrounding the nucleus.

Then, in 2007, Puglielli discovered a third location where proteins are acetylated, and “at that time, based on what was known, it was an impossible result,” he says.

The third location that Puglielli’s research identified as a site for acetylation was the endoplasmic reticulum or ER, a lacy network of tubes made of proteins and lipids that extend into the cytosol from the membrane surrounding the nucleus.

Puglielli’s research group would go on to discover how this machinery works and start to understand its biological functions.

Turns out, a major function of protein acetylation in the ER is facilitating the chatter that keeps our cells running smoothly.

“How does my right hand know what my left hand is doing?” asks Puglielli. “My brain is coordinating both of them, of course, through a network of nerves.”

But there are no nerves inside the billions of cells that make up our bodies. How do the tiny organelles within these cells—from the mitochondria that generate energy to the ribosomes that manufacture proteins—know what the other organelles are up to? How can they all work together to keep the cell functioning and us alive?

That’s where acetylation of proteins comes in, says Puglielli. Acetylation can affect several features of proteins, including which other proteins they can interact with, and whether they are able to take part in biochemical reactions that allow cells to function.

“We discovered that protein acetylation in the ER is a vital part of

the signaling machinery that allows the organelles in our cells to talk to each other,” says Puglielli. Mutations that disrupt protein acetylation can interrupt the cross-talk between cell organelles, which may lead to several different disorders.

Some of these disorders, like Alzheimer’s disease, may take decades to develop, because other biological processes may partially compensate for the disrupted process. Others disorders, such as autism, may manifest soon after birth.

*“When you make a new discovery that could lead you to help people, the feeling is incomparable”*

Puglielli is searching for novel therapies for the prevention and treatment of the neurodevelopmental and neurodegenerative disorders associated with dysfunctional ER acetylation of proteins.

These unknowns fuel Puglielli’s research. “Science gives you the excitement of a quest,” says Puglielli. “You are always out there on a big ocean, and you have to deal with the winds and the waves as you search for new lands. But when you spot an undiscovered island, when you make a new discovery that could lead you to help people, the feeling is incomparable.”

For the unabridged story please visit: [waisman.wisc.edu/newsletter.htm](http://waisman.wisc.edu/newsletter.htm)



# Waisman Center

## In the News



The Waisman Center **Autism Treatment Programs** were featured in a **Wisconsin State Journal** article about increasing options in the Madison area for children with autism and their families.

**Molly Murphy**, lead supervisor of the Autism Treatment Programs, and **Erin Thomson**, a clinical social worker with the programs, were part of the story.

You can read the story at [tinyurl.com/waismanATP](http://tinyurl.com/waismanATP)

Get to know Waisman Center Director **Albee Messing**! He was featured in the **Know your Madisonian** column in the **Wisconsin State Journal**.

Messing focuses on Alexander disease, a rare and usually fatal disorder of the nervous system. He is married to Marsha Mailick, vice chancellor for research and graduate education at UW-Madison and former director of the Waisman Center. They have four adult children and three grandchildren.

You can read the story at [tinyurl.com/waismanMessing](http://tinyurl.com/waismanMessing)



Waisman Center investigator **Seth Pollak's** research was recently highlighted in the American Psychological Association's **Monitor on Psychology** magazine.

Research projects in Pollak's lab are focused on children's emotional development and the relationship between early emotional experience and child psychopathology.

You can read the story at [tinyurl.com/SethPollak](http://tinyurl.com/SethPollak)

**Madison.com** reported that **Carl Ross** has been selected to serve as managing director of **Waisman Biomanufacturing**. His appointment started July 1, following more than a year as its interim director.

Located at the Waisman Center, the primary mission of Waisman Biomanufacturing is to help UW scientists and others efficiently translate scientific discoveries into early-stage clinical trials.

You can read the story at [tinyurl.com/waismanbio](http://tinyurl.com/waismanbio)





# UPCOMING EVENTS



## Waisman Center Children's Theatre

### 2017-2018 Schedule

All shows are on the **second Sunday** of each month

- ◆ Sunday, Oct. 8 @ 1:00 Ken Lonquist – fall songs & family tunes
- ◆ Sunday, Nov. 12 @ 1:00 & 3:00 Playtime Productions – *The Little Match Girl*  
*\*Sensory-friendly performance at 3:00 in partnership with the Autism Society of South Central Wisconsin*
- ◆ Sunday, Dec. 10 @ 1:00 & 3:00 Dance Wisconsin – *Nutcracker* preview
- ◆ Sunday, Jan. 14 @ 1:00 Wayne the Wizard – winter magic show
- ◆ Sunday, Feb. 11 @ 1:00 Mad Science – Spin, Pop, Boom!
- ◆ Sunday, Mar. 11 @ 1:00 Trinity Irish Dance – Irish music & dance
- ◆ Sunday, Apr. 8 @ 1:00 & 3:00 Playtime Productions – *The Reluctant Dragon*  
*\*Sensory-friendly performance at 3:00 in partnership with the Autism Society of South Central Wisconsin*

**Admission:** \$2.00, adults; \$1.00, children  
**Tickets:** Advance tickets for any show in the series can be purchased beginning at NOON on performance days  
**Sign Interpreters:** All shows sign-interpreted  
**Location:** Friends of the Waisman Center Auditorium, 1500 Highland Ave.  
**Directions:** [waisman.wisc.edu/wc-map.htm](http://waisman.wisc.edu/wc-map.htm)  
**Parking:** Free. Park in far end of lot 82, **overflow parking** in lots 60 and 76  
**Questions?** 608.263.5837 weekdays or [palumbo@waisman.wisc.edu](mailto:palumbo@waisman.wisc.edu)

Series sponsored by



Join us for early bird activities beginning at noon on the specially marked show dates. Interactive! Educational! Fun!

Series hosted by the Friends of the Waisman Center

[waisman.wisc.edu/events-ct.htm](http://waisman.wisc.edu/events-ct.htm)

## Waisman Center Day with the Experts

### Cerebral Palsy

**SATURDAY, Nov. 4, 2017 at 9 a.m.**



[waisman.wisc.edu/cerebral-palsy-2017.htm](http://waisman.wisc.edu/cerebral-palsy-2017.htm)

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The Friends of the Waisman Center



# Waisman Whirl

*Run, Walk & Roll for ALL Abilities*

3<sup>rd</sup> Annual

**Sunday, October 15, 10:00 a.m.**

Start and finish at the Waisman Center, 1500 Highland Ave.

**10k Run & Roll**

**5k Run, Walk & Roll**

**1 Mile Walk & Roll**

**Kids' Fun Run & Roll**

[waisman.wisc.edu/run2017.htm](http://waisman.wisc.edu/run2017.htm)

*Proceeds benefit individuals and families affected by developmental disabilities and neurodegenerative diseases.*



**Help pave the way for  
discovery and hope!**



**Please give at:**

[waisman.wisc.edu/giving.htm](http://waisman.wisc.edu/giving.htm)





## A SOFT Conference

Kari, Leila, James and Madelyn Adamson (left to right)  
Photo by Niamh Rahman, Waisman Communications

**When Leila Adamson** was born, doctors weren't sure that she would live for even 63 seconds.

More than 63 months later, in July 2017, she was in Madison, Wisconsin, along with her parents, Kari and James, and her sister, Madelyn, for the 31<sup>st</sup> annual conference of the Support Organization for Trisomy 18, 13 and Related Disorders, or SOFT.

Set up in 1980, SOFT provides "support and understanding to families...surrounding the diagnosis and care in trisomy 18, 13 and other related chromosomal disorders."

Leila has trisomy 18. Most of us have two copies of each of our 23 chromosomes—one set inherited from our father, the other from our mother—in our cells. Leila's cells, however, have three copies of chromosome 18, which can lead to high incidences of infant mortality and medical complications, like congenital heart problems and developmental challenges.

Trisomy 18, trisomy 13, and related disorders are all relatively rare; trisomy 18, for instance, occurs about once every 7,000 live births in the US. Their rarity can make it difficult for patient families to access effective medical care and meet other families facing similar challenges.

SOFT conferences try to address these challenges by connecting parents and families of children diagnosed with these genetic conditions and partnering with various medical centers and health care professionals, says **John Carey**, professor and vice chair of academic affairs in the Department of Pediatrics at the University of Utah, and a co-founder of SOFT.

The clinical aspect of this year's conference took place at the Waisman Center, in partnership with UW Health and the American Family Children's Hospital.

The Waisman Center has 11 specialty clinics and a long history of research as well as providing care and services to individuals with developmental challenges. Clinicians, including cardiologists, neurologists, geneticists, and developmental pediatricians, and LEND program trainees volunteered their time to meet with the families and offer suggestions and ideas.

"It was fantastic to be able to speak with specialists, maybe even get a second opinion, and discuss different treatment options for Leila," says Kari.

The conference also provided a great opportunity for the volunteers to meet and interact with families.

"It was a wonderful experience and a powerful reinforcement of the fact that we learn the most important lessons from our patients," says **David Wargowski**, a clinical biochemical geneticist at the Waisman Center Clinics.

For the Adamsons, "it was very exciting to see the older kids, and also fun to snuggle the young babies because we got to see how far Leila has come," says Kari.

And Leila has come a long way. "She likes to jump and spin, she likes to spend time in her little sensory room with lights, and she loves cartoons," says James. At the mention of cartoons, Leila, who is non-verbal, squeals in delight. Her joy is evident.

The conference also included several other sessions, such as workshops on the legal aspects of special-needs trusts and guardianship, CPR training, and communication using iPads and computers.

Carey is proud that the conference combines social, medical and informational support, which are all core elements of SOFT's mission. "Ultimately, it's all about the patients and their families," he says.

*To read the unabridged story, please visit:*  
[waisman.wisc.edu/newsletter.htm](http://waisman.wisc.edu/newsletter.htm)





1500 Highland Avenue, Madison, WI 53705

Director's Office: 608.263.5940

[waisman.wisc.edu](http://waisman.wisc.edu)



## Art for All

Encompassing a breadth of subject matter and artistic media, the **Harvey A. Stevens International Collection of Art by People with Developmental Disabilities** showcases unique and visually powerful pieces that encourage people of all abilities to express themselves and expand their world through art.

Our recent Call for Art garnered a tremendous response, with more than 75 beautiful submissions from across the US and the world, including the work pictured here!

For more information, please contact **Teresa Palumbo** at [palumbo@waisman.wisc.edu](mailto:palumbo@waisman.wisc.edu) or 608.263.5837

*Swimming with the Fishes*,  
by Alex Scott, an artist from  
Chicago.