



Clinic for  
Special Children

# Newsletter

ISSUE #44

SUMMER 2018



## "Darren Lee is an Angel Among Us" The Martin Family

"An angel among us," is how Ruth Martin describes her son, Darren Lee Martin ('Lee'). Pictured above with his parents, Ruth and Darren, and older brothers Adam and Tristan, Lee's joy is evident.

Born in a hospital setting, there was nothing notable about Lee's birth. But at six weeks old, mom and dad started to notice that Lee wouldn't giggle, laugh, and was unable to follow them with his eyes. After several visits to pediatricians and specialists, the Martin family was referred to Clinic for Special Children (CSC).

Lee first visited CSC when he was six months old. Dr. Strauss worked to manage Lee's symptoms while Dr. Puffenberger worked towards a genetic diagnosis. When CSC started collaborating with the Regeneron Genetic Center in 2014, the Martin's DNA was one of the first sent for an advanced genetic testing called exome sequencing. A mutation was found in one of the five genes known to cause Kleefstra Syndrome (KS). Like most children with KS, Lee had a de novo mutation, meaning the genetic change causing his symptoms was not inherited from either parent.

Kleefstra Syndrome is characterized by intellectual disability and a spectrum of physical and clinical symptoms. Since the symptoms can be diverse, it makes managing care challenging.

Although there are no cures for KS, there are many options for the management of symptoms.

"Having a special child has changed our outlook on life. Most importantly, we are committed to live life as normal as possible and allow Lee to experience the same joys as other children." Since there is no definitive prognosis for Lee, the Martins face one day at a time. Darren and Ruth have found strength in their faith and support from their family and friends.

Now a vibrant nine-year-old boy, Lee's favorite things are to wrestle with his big brothers, spend time outdoors, take car rides, and swinging.

"We are so grateful to Dr. Strauss and the Clinic team for their compassion. Dr. Strauss has made a big difference for us, always making sure we, his family, are also taken care of as well as Lee," says Ruth.

"Life doesn't stop when your child is diagnosed with a rare disorder." To parents living with special children, Ruth offers the advice, "Through the roller coaster of emotions you may experience, give yourself time. Wherever you are in your journey, it's OK to be where you are and feel the way you do. Everybody's story is different and we wanted to tell ours in case we could inspire one person."



## SPECIAL DATES

### Missouri Benefit Auction Saturday, August 18

Ed's Machinery LLC  
17920 US-136  
Memphis, MO 63555  
9:30 a.m. - 4 p.m.

### Blair County Benefit Auction Saturday, September 8

Morrison's Cove Produce Auction  
174 Windy Acres Ln.  
Roaring Spring, PA 16673  
7 a.m. - 4 p.m.

### Patient Family Advisory Council Meeting

Tuesday, September 11  
The Clinic, 7 p.m. - 8 p.m.

### SMA Family Day Wednesday, September 19

The Clinic, 9 a.m. - 5 p.m.

### CSC 1st Annual 5K RUN/WALK Saturday, September 22

The Clinic, 8 a.m. - 10 a.m. - Application inside

### Patient Fall Family Fun Day Saturday, October 13

The Clinic, 12 p.m. - 4 p.m. - Details inside

### 2018 Extraordinary Give Friday, November 16

Online Fundraiser  
[www.ExtraGive.org](http://www.ExtraGive.org)  
12:01 a.m. - 11:59 p.m.

Call the office, check our Facebook page, or our website for event specific information and updates.



## Staff News

### Kelly Cullen



Kelly joined the Clinic for Special Children in July as the Communications Manager. In this role, Kelly manages the Clinic's newsletter, event promotion, social media, and website. She earned her Bachelor's of Science in Business Administration/Marketing from Elizabethtown College in 2013. Prior to joining the Clinic, Kelly was the Global Creative Brand Manager for a baby gear company where she was involved in all facets of brand marketing for the organization.

"As soon as I learned about the Clinic, I knew I wanted to be a part of this team that's at the forefront of genomic research and treatment. It is an honor to work in a role where I can help others as well as use my experiences to further the Clinic's mission."

### Lauren Bowser



In the summer of 2017, Lauren spent three months at Clinic for Special Children as a student researcher working on a Spinal Muscular Atrophy (SMA) natural history study. As a student at Franklin & Marshall College, she continued the research in Fall 2017 through an independent study project. In Spring 2018, she began working on a second independent study project, this time on the natural history of GM3 synthase deficiency. Lauren graduated in May 2018 with a Bachelor of Arts degree in neuroscience.

Following graduation, Lauren joined the Clinic as a full-time Research Fellow. In this position she is responsible for assisting with research efforts through data collection, data entry and management, data analysis, and manuscript preparation. "Service to others, particularly children, is what I intend to devote my life to as a future physician and the Clinic is a wonderful place to get started. I am continuously inspired by the generous spirits and kind hearts of the community as they pour endless love over their special children."

## New Position at CSC

### Kim Calderwood

Kim joined the Clinic for Special Children in 2016 as the Clinic's Communications Manager. In June she transitioned to a part-time Special Projects Assistant role. In this role she provides support to events and administrative functions at the Clinic. She also coordinates credentialing for CSC's clinical staff. "I'm very grateful for this opportunity to serve at the Clinic. My new role will allow me to continue to support the mission of the Clinic as I begin my new journey as a stay-at-home mom."



## Recently Published Paper

### Amish Nemaline Myopathy Natural History Study Finds Promise for Gene Therapy Treatment

Drs. Carson, Strauss and Fox and Karlla Brigatti were part of a team that recently published a natural history study paper about Amish Nemaline Myopathy (ANM) in the Old Order Amish population and the promise of gene therapy for the lethal disorder. The study summarizes genealogical records, clinical data, and molecular reports of one hundred and six ANM patients born between 1923 and 2017. It appeared this month in the journal *Human Molecular Genetics*.

All ANM patients in the study were born at a normal birth weight, failed to thrive by 9 months of age, and died at a median age of 18 months from respiratory failure. Common symptoms of ANM shortly after birth include low muscle tone, hip and shoulder stiffness, and tremors, followed by progressive muscle weakness, degeneration and joint contractures.

Muscle biopsies from two ANM patients showed an abnormal pattern of muscle fibers with preserved nerve function. Researchers compared the human ANM muscle biopsies with those from a transgenic mouse model; they were very similar, suggesting the mice can serve as a good model of the human disease. At the Clinic for Special Children, genetic testing allows practitioners to confirm the diagnosis of ANM within days of birth. The findings in this study provide a strong platform for exploring gene replacement therapy in newborns diagnosed with ANM.

## Patient Fall Family Fun Day!

### Hosted by the Clinic's Patient Family Advisory Council

**Saturday, October 13**

**12 p.m. - 4 p.m.**

**at Clinic for Special Children**

Patients and their families are invited to join the Clinic's staff and board for a FREE day of fellowship and fun! Get to know the Clinic's staff, board, supporting specialists, and their families! Enjoy activities at the Clinic between 12 p.m. and 4 p.m.: hayrides with antique tractors, a tethered hot air balloon, children's activities, tours of the Clinic, delicious food, fresh cider, ice cream, and more!

The event is hosted by the Patient Family Advisory Council (PFAC). The PFAC is a group of patients, family members, clinicians, and staff members that meet bimonthly with the purpose of incorporating a patient voice into the Clinic's programs and practices.



## Clinic for Special Children

### 5K Run/Walk/Jog

**Saturday, September 22**

**8 a.m. - 10 a.m.**

**at the Clinic for Special Children**

Please join us for our inaugural 5K to benefit the Clinic for Special Children! All race proceeds support the mission of the Clinic.

Runners, joggers, and walkers are invited to join us for a 3.1 mile easy course starting and ending at the Clinic for Special Children in Strasburg Township along scenic country roads, winding through Lancaster County farmland. Participants will have full use of the closed road as they venture past vineyards, see an Amish schoolhouse, and pass by acres of picturesque Amish farms. A cash prize of \$100 will be awarded to top overall male and female runners!

### A Family Friendly Event:

**Crafts, baked goods sale, silent  
auction, quilts, free refreshments!**

### Register by 9/1 to receive a discount

Mail the application in this newsletter to CSC in the envelope or visit [clinicforspecialchildren.org](http://clinicforspecialchildren.org) to register. Call CSC if you need more applications.



## Help for Family from Remote Village in Mexico

# Margaretha

While many families often travel considerable distance to receive medical care, four-year-old Margaretha and her parents made a nearly 2,500 mile journey from southeast Mexico to Delaware to do so. They are Old Colony Mennonites who live in the Campeche region of Mexico, descended from Prussian Mennonite colonists who migrated from Russia to settlements in Canada in the 1870s, and then into Mexico a few generations later. They speak Plautdietsch, or Mennonite Low German (and some Spanish) and are faithful to the traditions and lifestyles of their Mennonite ancestors. On several occasions arrangements were made for a Plaut-speaker to serve as an intermediary to ensure the family understood all of the medical team's explanations.

Dr. Strauss and Karlla Brigatti met Margaretha when they traveled to the colony last August with Clinic for Special Children Board of Directors chairman Herman Bontrager, members from Mexico Mennonite Aid (MMA), and the Anabaptist Foundation on a medical mission. Several families there have children with special needs, and so Dr. Strauss and Karlla met with over 30 patients for several days and devised care plans that could be implemented locally.

However, some children had serious medical problems that could not be managed in Mexico, including Margaretha. She was born with cloacal extrophy, a very rare and severe birth defect where most of the abdominal organs, including the bladder, intestines, and spinal cord, are externalized and exposed early during development in the womb, causing multiple serious health issues after birth. Margaretha was able to have some of the necessary surgeries in Mexico, but she still required constant care and remained at risk for life-threatening complications. Despite these challenges, she was a lively, happy child, and her family was very dedicated to her care.

Once back home, Dr. Strauss worked closely with International Medicine at the Nemours/ Alfred I. duPont Hospital for Children, the Anabaptist Foundation, and MMA to determine the feasibility and logistics for Margaretha to have surgery at Nemours. At long last, Margaretha and her parents were able to come to Nemours in early June. While awaiting her surgery, she was able to visit with cousins in Canada and see Washington, DC and Virginia as well. Her surgeries took over 14 hours, but she made a full recovery and will have a much better quality of life as a result. She also enjoyed her stay at the Ronald McDonald House at Nemours and the family found everyone at the hospital incredibly helpful and kind. When Dr. Strauss and Karlla went to Nemours to see her in late July, Margaretha's parents were looking forward to returning to Mexico and resuming their normal lives, but happy they made the trip and very pleased with Margaretha's care. They were grateful to everyone who made that possible and everyone's good wishes for her recovery. The joint efforts from the Clinic for Special Children, MMA, the Anabaptist Foundation, and Nemours made a big impact on a very special little girl!

## Mark your calendar for the remaining 2018 Benefit Auctions

Please join us for days of fellowship, family, and food! Handmade quilts, handcrafted furniture, wooden crafts, sporting goods, handmade toys, and garden plants will be available to bidders. A variety of food choices will be featured including chicken barbecue, freshly made pies, donuts, whoopie pies, ice cream, and more!

### Missouri Benefit Auction - 8/18/18

9:30 a.m. Candy Drop | 10:15 a.m. Auction Begins

Lunch to Follow at 11:00 a.m.

Ed's Machinery LLC

17920 US-136

Memphis, MO 63555

Contact: Harlan Burkholder | 660-341-4113

### Blair County Auction - 9/8/18

7:00 a.m. Breakfast | 8:30 a.m. Auction Begins

Physician Remarks and Quilts to Follow

Morrison's Cove Produce

174 Windy Acres Ln.

Roaring Spring, PA 16673

Contact: Paul Ray Fox | 814-224-5442

Making up approximately 50% of the Clinic's revenue, funds raised from our annual benefit auctions and charitable donations make it possible for the Clinic to continue to provide affordable, specialized care to children living with rare genetic disorders not just in Lancaster County, but across the country and world.

## Giving Opportunity: Local Family Foundation Match

Thanks to the generosity of a local family foundation, we are excited to announce a \$15,000, 1-for-1 match to your donations that expires on December 3, 2018! Beginning now, your \$100 gift becomes \$200, your \$250 gift becomes \$500, your \$500 becomes \$1,000!

All gifts support the mission of Clinic for Special Children, a non-profit medical home for children living with rare disease.



[clinicforspecialchildren.org](http://clinicforspecialchildren.org)





Pictured from left to right: Dr. Matt Demczko and Dr. Mike Fox

## Returning to Clinic for Special Children Dr. Matt Demczko and Dr. Mike Fox

Last year, Clinic for Special Children (CSC) enjoyed the privilege of working closely with Nemours and two physicians who were leading efforts to open a new medical clinic in Dover, Delaware to serve the local Amish community. In preparation for opening the new Dover clinic, CSC agreed to host Drs. Fox and Demczko for six month fellowships, giving them the unique opportunity to learn how to care for Amish and Mennonite children living with rare genetic disorders.

Since their previous work with CSC, the new Kinder Clinic has opened which is the first of its kind to offer service directed toward children with special needs in southern Delaware from the Plain Communities. The Kinder Clinic is owned and operated by Nemours, a non-profit pediatric health system. Over time, it is the hope of the clinical team to also provide patients access to pediatric subspecialists close to home for their patients.

We are happy to announce that Drs. Fox and Demczko are temporarily returning to the CSC clinical team. Until CSC fills an open full-time physician position, we will be leasing the services of both physicians from Nemours part-time. Since July, the physicians have been working at CSC on Tuesdays and Thursdays. They are evaluating new patients and providing care to current patients.

Given their previous experience training at CSC, it allowed Drs. Fox and Demczko to jump right in and start seeing patients as few people could. We appreciate Nemours and Drs. Fox and Demczko's willingness to fill in until a new, full-time physician is identified. Their service is invaluable to CSC.

## Questions and Answers Carrier Testing

### What is carrier testing?

Carrier testing is an elective genetic test performed to determine if someone has a change in a specific gene that is known to cause a genetic condition. Common examples of these conditions include Maple Syrup Urine Disease (MSUD), Ellis-van Creveld Syndrome (EVC), Spinal Muscular Atrophy (SMA) and Glutaric Acidemia Type 1 (GA-1).

### Why is carrier testing important?

The goal of carrier testing is to detect couples who are at risk of having children with a genetic condition. This knowledge can truly make a difference for an affected newborn, such as immediate diagnosis and treatment after birth, if necessary. This leads to the best possible outcome for affected babies.

### Who should be tested?

Carrier testing is most useful for couples starting a family, before or during a pregnancy. Family history of a condition can determine which tests are most important to consider, though many carriers do not have a family history that would suggest specific testing. The Clinic for Special Children is developing a test for all genetic changes found in the Plain populations, optimizing detection of at-risk couples and their affected children.

### Should both parents be tested?

Most genetic conditions in the Plain communities are *recessive*, meaning the condition is only apparent when both copies of the gene in an individual have the alteration. Carrier parents each have one copy of the gene that functions properly, and a second copy with the alteration associated with the disease. Therefore knowing the carrier status of *both* parents is most informative.

### What if one parent is a carrier?

For recessive conditions, it would be extremely unlikely for a couple to have an affected baby when only one parent is a carrier for the condition. However, these children may be carriers as well, and should undergo testing in adulthood.

### Contact information and fees

Carrier testing involves a simple blood test and can be arranged in your area. Please contact your local midwife or the Clinic at 717-687-9407 for additional information or to schedule a test.

Carrier test (individual): \$50

Carrier test (couple): \$85

\*Shipping costs and fees for additional testing are not included.

[clinicforspecialchildren.org](http://clinicforspecialchildren.org)





Pictured from left to right: Dr. Kyle Soltys and Dr. George Mazariegos

**"I enjoy having the ability to connect something that is very personal or a difficult part of someone's journey with an innovative and technical field like transplant is."**

-Dr. Mazariegos

## Collaborator Spotlight: George Mazariegos, MD and Kyle Soltys, MD

### Children's Hospital of Pittsburgh Transplant Team

For over twenty years, the transplant team at the Children's Hospital of Pittsburgh (CHP) has provided life-saving organ transplants to patients of the Clinic for Special Children (CSC). Due to the metabolic nature of many disorders treated at CSC, Dr. George Mazariegos, Director of Pediatric Transplantation at CHP, and Dr. Kyle Soltys, Pediatric Liver Transplant Surgeon, have worked closely with CSC to arrange and perform liver transplants, working personally with many patients and families cared for by the Clinic for Special Children.

Dr. Mazariegos knew from a young age that he wanted to practice medicine. This desire was strengthened by an opportunity he took advantage of in high school, volunteering at a local hospital. Following high school graduation, he was accepted to a competitive accelerated schooling program through Northwestern University that included admission into their medical school after two years of undergraduate studies. Dr. Mazariegos completed his residency training through Michigan State University and his fellowship training through University of Pittsburgh.

"In 1989, the field of transplant was just developing in Pittsburgh." After Dr. Mazariegos completed his fellowship training, he joined the Thomas E. Starzl Transplantation Institute and in 1997 joined the Children's Hospital of Pittsburgh as Codirector of Pediatric Liver and Intestine Transplantation. When asked about his career, Dr. Mazariegos explains, "As a transplant surgeon, I enjoy having the ability to connect something that is a very personal or difficult part of someone's journey with an innovative and technical field like transplant is. This merging of the real challenges faced by families or patients with technology and the capacity that God has given us to serve patients and putting it together in that way is rewarding."

Dr. Soltys graduated from Boston University School of Medicine and performed his residency in general surgery at University Hospital in Newark, New Jersey. He also completed his initial fellowship in transplantation at Mount Sinai Medical Center in New York before heading to Pittsburgh as a fellow at the Thomas E. Starzl Transplant

Institute of UPMC in 2003. He has authored more than 70 published medical journal articles and is an Associate Professor of Surgery at the University of Pittsburgh Medical School.

Though CHP's transplant team began collaborating with CSC many years ago, in 2003-2004 the team at CSC and the transplant team from CHP laid the groundwork for the protocol for liver transplants for children living with Maple Syrup Urine Disease, specifically planning the transplant of patients from CSC. Over the course of time that CSC and CHP have worked together, over 100 CSC patients have been successfully transplanted. Each year Drs. Mazariegos and Soltys and the CHP team visit CSC for the annual Transplant Family Day where they give talks, conduct annual patient evaluations and meet with the CSC staff.

Dr. Mazariegos says in the near future he hopes the collaboration will lead to new, innovative therapies and alternative treatments for children living with rare genetic disease. We are grateful to the CHP team for their collaboration over the years and look forward to the years to come.



2018 Transplant Family Day at the Clinic including the CSC & CHP team





## Our Staff

Keturah Beiler, RN  
Nurse

Lauren Bowser  
Research Fellow

Karlla Brigatti, MS, LCGC  
Research Operations Director and  
Genetic Counselor

Kim Calderwood, MA  
Special Projects Assistant

Vincent Carson, MD  
Pediatric Neurologist

Kelly Cullen  
Communications Manager

Adam D. Heaps, MS, MBA  
Executive Director

Christine Hendrickson, RNC  
Nurse

Candace Kendig  
Medical Receptionist

Lavina King  
Community Liaison

Yalonda L. Kosek  
Office Coordinator

KaLynn Loeven  
Laboratory Technician

Erik G. Puffenberger, PhD  
Laboratory Director

Teresa Rineer  
Development Director

Donna L. Robinson, CRNP  
Nurse Practitioner

Ashlin Rodrigues  
Laboratory Technician

Kevin A. Strauss, MD  
Medical Director

Millie Young, RNC  
Research Nurse

## Board of Directors

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Chair-Charity Committee

Herman Bontrager  
Chairman

Richard Fluck, PhD  
Secretary, Chair-Development Committee

Leon Hoover

Leonard Hurst

Mark Martin  
Treasurer

Jacob Petersheim

Stephen D. Ratcliffe, MD, MSPH

Jacob Zook  
Vice Chairman



## Donor Spotlight

### Supporting the Clinic Stitch by Stitch

Jordan McCroskey, General Manager of Oklahoma Embroidery and Design (OESD), speaks with excitement when describing the creativity and fun behind embroidering. His business, OESD, was founded in Oklahoma in 1987 and creates beautiful embroidery designs for quilts, decorations, tiling scenes, and more! Many of his suppliers and networkers are from the Plain community all over the United States. When the chance arose earlier this year to support the Clinic for Special Children (CSC), the OESD team (pictured above at their annual holiday party) developed the “patchwork” for an original fundraiser.

Jordan first learned of CSC when his long-time business colleague and friend, Leon Hoover, approached him about supporting the annual Clinic for Special Children auction in Union County. Leon is a board member of CSC, organizer of the Union County Auction, and owner of Hoover’s Bernina Sew in Mifflinburg, PA. After speaking with Leon, the OESD team knew they wanted to contribute in a big way and started generating unique ideas to contribute to the CSC’s mission.

When asked why the OESD team decided to support CSC, Jordan’s reply was simple, “Dr. Kevin Strauss’s TEDx talk.” Jordan and his staff collectively watch TED and TEDx talks frequently to generate innovative ideas for their business, elevate their customer service and keep up-to-date on learning opportunities. When the OESD staff had to pick a charity that would receive proceeds from a limited-time embroidery collection, they knew they found their fit after watching Dr. Kevin Strauss’s 2014 TEDx talk, “Decoding the Unique Medical Needs of the Amish.” Jordan saw this fundraising opportunity as a perfect way to support the Plain community as well as use his team’s expertise in embroidery design.

Earlier in the year, OESD put together a limited-time embroidery collection featuring 12 staff-favorite designs. Sales of the collection generated over \$2,000 in proceeds for CSC! Additionally, their support demonstrates the growth of CSC’s mission and the various creative ways that community members, all over the globe, support CSC. Thank you to the Oklahoma Embroidery and Design for their support!



# Patient Fall Family Fun Day

October 13, 2018  
12 p.m.-4 p.m.

The Clinic for Special Children's Patient Family Advisory Council  
invites patients and their families to CSC's second annual

## Fall Family Fun Day

Clinic for Special Children  
535 Bunker Hill Road | Strasburg, PA  
October 13th 12pm - 4pm

### Family Activities!

Hayrides with Antique Tractors - Kid's Activities - Tethered Hot Air Balloon

### Delicious Food!

Hot Dogs - Fresh Cut French Fries - Rita's Ice - Local Apples and more!



You and your family are invited to join the CSC staff for a FREE day of fellowship and fun! Get to know the CSC staff, physicians, supporting specialists, and their families! Enjoy activities at the Clinic between 12pm and 4pm. Refreshments will be provided for you.

Please call us and let us know you're coming! 717-687-9407

This event is hosted by CSC's Patient Family Advisory Council, a collaborative group of patients, family members, clinicians, and staff members that meet bi-monthly with the purpose of adding a patient voice to the Clinic's programs and practices.

 Clinic for Special Children





# Clinic for Special Children

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## The Clinic's Mission

"To serve children and adults who suffer from genetic and other complex medical disorders by providing comprehensive medical, laboratory, and consultive services, and by increasing and disseminating knowledge of science and medicine."

Charles H. Winters, Jr.  
To 50 R.R. Harris 300 90