



Clinic for
Special Children

Newsletter

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A Family's Journey with GA-1 The Watson Family

"The Clinic for Special Children is a shining star on many people's journeys and I hope more people can find that star." Ruth Watson speaks of frustrations, struggles and ultimately peace when describing her family's journey in diagnosing and treating her son, Chris. Ruth had a routine pregnancy, delivery and first several months with her newborn son in the mid-1990s. However, when Chris was nine months old, she got a call from the daycare that would change her family's lives. The daycare told Ruth that Chris had a cold and wasn't breathing well; when Ruth picked him up, she says she was handed "a gray, limp child."

This was the first of many visits to the hospital for Ruth and her family. The physicians couldn't find anything wrong and sent the family home. Several days after this initial hospital visit, her husband Bill called and said that Chris had a seizure and was rushed to the local Emergency Room.

After speaking with a pediatrician, they were released to another hospital in the area. Many of Chris' symptoms pointed the physicians to diagnose Shaken Baby Syndrome which then started many rounds of police questioning for Ruth and Bill, the daycare, grandparents and anyone with direct contact with Chris.

In 1997, they met with another doctor who said he didn't believe the previous diagnoses. After testing and consulting with a team of medical students, Ruth and Bill finally got a diagnosis - Glutaric Acidemia Type 1 (GA-1).

Ruth and Bill poured themselves into researching GA-1, which in 1997, the information for patient expectancy was dismal at best. Through this research they found Dr. Morton's information and called the Clinic for Special Children. Ruth describes after their first call with Dr. Morton, "we knew we found the guy." They used all of the formula as suggested by the Clinic and saw great improvements in Chris' condition.

In September 1998, the Watson's were able to visit the Clinic in Strasburg. During this trip they attended a local GA-1 meeting at a local restaurant, and Ruth was shocked at how well Chris' condition was compared to other children in the group. At this moment she realized he truly was one of "the lucky ones."

Chris Watson is now a senior at the University of Denver studying Psychology with a minor in Computer Science and Communications. He plans to head home for a year to intern and then go to Texas A&M for a Master's Degree. The story of Chris Watson is a true testament of the Clinic's work of impacting lives in childhood and beyond.



SPECIAL DATES

The Great Train Robbery Saturday, October 20

Strasburg Rail Road

www.StrasburgRailroad.com

4 p.m.-5 p.m. (Event donations support CSC)

2018 Extraordinary Give Friday, November 16

Online Fundraiser, All Day - Details inside

Thanksgiving - Office Closed November 22 & 23

All Day

The Night Before Christmas Train November 30 - December 21

Strasburg Rail Road

www.StrasburgRailroad.com

Portion of ticket sales benefits CSC

Christmas Eve - Office Closed Monday, December 24

1 p.m.-5 p.m.

Christmas Holiday - Office Closed December 25 & 26

All Day

New Years Eve - Office Closed Monday, December 31

1 p.m.-5 p.m.

New Years Day - Office Closed Tuesday, January 1

All Day

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

We're Growing! New Positions at the Clinic

Julia Martin



Julia Martin is the mother of a child living with Maple Syrup Urine Disease (MSUD), cared for by the Clinic for Special Children. Over the past year, Julia and her husband, Daryl have been instrumental in helping to establish the Clinic's

Patient Family Advisory Council (PFAC). In collaboration with the PFAC families, Julia helped to coordinate and organize the Clinic's first Fall Family Fun Day for patients and their families. In September, Julia officially joined the Clinic for Special Children's team as our first Development Assistant and will be working two days a week. She will work with our Development Director, Teresa, to ensure an excellent experience for the Clinic's donors and stakeholders.

Emily Seitz

Emily joined the Clinic for Special Children in August as a part-time Scientific Grant Writer. Emily is



currently a PhD candidate at the Pennsylvania State University in the Departments of History and Women's, Gender, and Sexuality Studies. Her dissertation is titled "What

About the Mother?: Managing Maternal Mortality in Philadelphia, 1850-1973." She plans to defend her dissertation in the Spring of 2019. She has a MA in History and Women's, Gender, and Sexuality Studies from Pennsylvania State University and a BA in History and Behavioral Science from York College. Emily is also the Assistant Director for Fellowships and Grants in the humanities and social sciences at Franklin & Marshall College.

It's Flu Shot Season

Please call the office at 717-687-9407 to schedule your flu shot. Anyone over six months of age is encouraged to get the flu shot. We offer shots to our patients and their immediate family for \$20 per person. The best way to protect yourself and your family is to avoid exposure, practice good handwashing, and receive an annual flu shot.

Recently Published Papers

Spinal Muscular Atrophy (SMA) is a genetic condition that causes progressive degeneration of the motor nerve cells in the spinal cord and brainstem resulting in weakness of the muscles involved in breathing, eating and voluntary movement. It is the most common genetic cause of infant death worldwide and is found in the Plain communities. The severity of the disease is determined by genetic factors and ranges from the most common severe form (type 1) which is typically lethal in childhood, to less severe, adult onset forms (type 4).

The staff at the Clinic have cared for a number of patients with SMA over the years and are unfortunately well aware of the toll it takes on families and communities. Until recently, most treatment was focused on managing the symptoms of the disease. A new pharmaceutical therapy, Nusinersen, also called SPINRAZA®, was approved in late 2016 and clinical trials are on-going for gene replacement therapy. In newborns who do not yet show symptoms of the condition, the Clinic for Special Children is a site for both therapeutic approaches. Over the last year, the Clinic staff has been working hard to bring innovative solutions to SMA families in practical and affordable ways. Recently Clinic staff published two papers related to SMA: a natural history study summarizing the SMA experience in the Plain communities and an initial report of a new drug delivery method pioneered by the Clinic and A.I. duPont Hospital for Children/Nemours.

Preliminary safety and tolerability of a novel subcutaneous intrathecal catheter system for repeated outpatient dosing of nusinersen to children and adults with SMA.

Nusinersen, also called SPINRAZA®, was approved in November 2016 to treat SMA. The drug has to be injected into the fluid around the spinal cord, called cerebral spinal fluid (CSF), every four months after the initial loading doses. Typically the drug is injected into the CSF by a lumbar puncture, commonly known as a spinal tap. However, many older SMA patients have problems with their spine related to their disease which makes it very difficult to perform a lumbar puncture safely.

In order to be able to administer the medication to these patients, the Clinic for Special Children worked with A.I. duPont Hospital for Children to develop a catheter system to deliver the drug intrathecally. This

means that an implanted catheter is placed in the CSF and is connected to an infusion port that sits under the skin. The drug can then be given by injecting it with a needle into the port, rather than having to perform a lumbar puncture. This is a procedure that can be done in an outpatient setting rather than in a hospital.

The paper summarized the Clinic's experience in the first ten patients implanted with this device, who all tolerated the surgery well. Overall, the cost of drug administration was significantly reduced compared to repeated lumbar punctures. This is a method that is of significance not just for patients that have spinal issues, but anyone who would be receiving the drug. The study so far has shown the catheter devices to be relatively safe and warrants further trials¹. If proven effective, this method could double the number of patients able to receive nusinersen worldwide while reducing costs five-to-ten fold.

¹This study is ongoing at the Clinic and open to new participants.

SMA within Amish and Mennonite populations: Ancestral haplotypes and natural history.

Natural history studies are a collection of patient experiences with a specific disorder over time, summarized into one report, which provides a comprehensive clinical description of the disease course. This can serve as the baseline to which new interventions are compared.

In this paper, the natural history of SMA is presented using information from forty-two Mennonite and fourteen Amish patients. It also outlines how certain haplotypes (pieces of DNA with certain genetic markers that are inherited together) predict disease severity. In Mennonite and Amish patients, three SMA haplotypes and one SMA haplotype were identified respectively. Mennonite patients were observed to either have SMA type 1 (patients typically never roll or sit) or SMA type 2 (most sit, some walk). All Amish patients were observed to have SMA type 1.

For children with SMA type 1, despite having similar genetic markers, had important differences in onset and severity of symptoms. Both groups tended to have a more severe course of disease than patients from other backgrounds, with Amish patients having the most decreased survival for unknown reasons. More work is needed to understand how the course of disease is affected by genetics or other factors.

2018 Auction Recap

Thank you to EVERYONE who made our 2018 auction season such a success! Over 50% of the Clinic's revenue comes from the annual auctions and charitable donations. We appreciate all of the committee members, volunteers, donors, bidders, and all supporters who make our auctions come to life!

Please visit our website (www.ClinicforSpecialChildren.org) to view the most current auction information for the upcoming 2019 season.



2018 Extraordinary Give

THE EXTRA ORDINARY GIVE



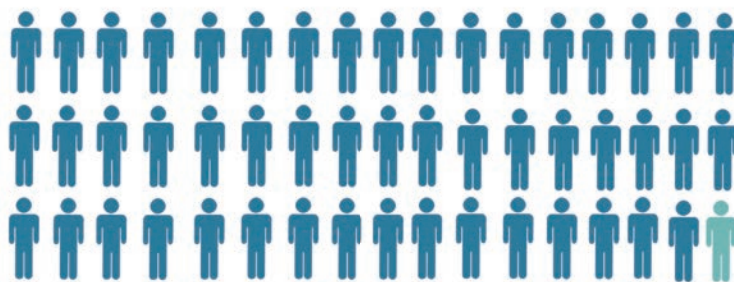
Friday, November 16
Midnight - 11:59 p.m.
Online Giving at ExtraGive.org

It's Lancaster County's largest day of online giving benefiting more than 400 local non-profit organizations! Every dollar donated through ExtraGive.org on November 16 will be stretched by a pool of more than \$500,000 from the Lancaster County Community Foundation, Rodgers & Associates, the High Foundation and other supporters.

Donating is simple. On Friday, November 16 visit ExtraGive.org and select 'Clinic for Special Children' as your non-profit organization of choice. If you would prefer to donate over the phone, please call the Clinic on November 16 between the hours of 9 a.m. - 5 p.m. and we would be happy to receive your gift over the phone.

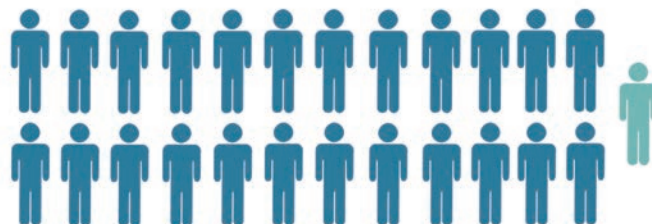
Visit us on November 16 from 11 a.m. to 3 p.m. in Lancaster City's Penn Square for our annual Whoopie Pie Toss in celebration of the Extraordinary Give! We appreciate your support!





1 in 50
people worldwide is
a carrier for SMA

SMA Carrier Frequencies



1 in 25
Mennonites is a carrier for SMA



1 in 8
Hutterites is a carrier for SMA

SMA Carrier Testing Now Available

Free of Cost

The Clinic for Special Children is excited to launch the SMA Prevention Readiness program, **offering genetic testing for spinal muscular atrophy (SMA) free of charge** to adults from the Plain community. The goal of this program is to identify individuals and couples who are carriers for this condition, to present therapeutic options to at-risk couples should they have an affected child, and to ensure that any newborn with SMA in the Plain community is diagnosed within the first few days of life, before the onset of symptoms.

SMA is a progressive genetic condition affecting the motor nerve cells (neurons) in the spinal cord and brainstem that affect movement, breathing, and eating. It is the most common genetic cause of infant death worldwide and is found in the Plain communities as well. However, recent targeted therapies have changed the course of this condition and improved the quality of life for affected individuals everywhere. Studies have shown that the earlier these treatments are given, the better they work. For this reason, we hope to give parents of affected children the option to begin their treatment of choice as soon as they can. Unfortunately, this is not a condition yet detected through newborn screening (the heel-prick test) in Pennsylvania.

We estimate that 1 in 25 Mennonites is a carrier for SMA, making it one of the more common conditions in the Mennonite community. We don't know the carrier frequency in the Amish, but several babies with SMA have been diagnosed within Amish communities across the country. Since carriers don't often have a family history of SMA, **we encourage all couples to consider carrier testing for this condition, even if they already have healthy children and no affected relatives.** If couples are considering carrier testing for other common conditions like MSUD or GA1, we can do SMA testing from the same sample. Like our other genetic testing, a healthcare provider such as a midwife or doctor can send this to us, and we always welcome those who want to come to the Clinic itself for testing. We'll follow-up after results are available to ensure they are clear.

Please feel free to contact Karlla Brigatti, Millie Young, or Lauren Bowser at the Clinic at 717-687-9407 to learn more about the SMA Prevention Readiness program, learn about new SMA therapies, or arrange for carrier testing.



Dr. Stephen Ratcliffe



Dr. Jiefu Yuan



Dr. Grace Loudon

Collaborator Spotlight: Lancaster General Health Family Medicine Residency

Dr. Stephen Ratcliffe with residents Dr. Jiefu Yuan and Dr. Grace Loudon

For 50 years, the Lancaster General Health (LGH) Family Medicine program has trained hundreds of family medicine residents in Lancaster, PA. Dr. Stephen Ratcliffe, Program Director of the Family Medicine residency and Clinic for Special Children board member, has been a vital component of the program's success. It has been the top-rated Family Medicine residency program on the East Coast for the past four consecutive years.

The Clinic first became involved with the residency program in the summer of 2015, when the program began providing inpatient care for adult patients when they were admitted to Lancaster General Hospital. During that same year, LGH family medicine residents began to do two to four week clinical rotations at the Clinic. In 2017, an Area of Concentration in primary care genomics was established at the Clinic to provide training to residents in clinical genomics in the care of adults and children.

Dr. Ratcliffe explains, "Our program has 17 Areas of Concentration (AOC); each of them offers a family medicine resident the opportunity to spend at least 200 hours of work in a given clinical area that occurs in addition to the extensive inpatient and outpatient training that our residents undergo throughout the three years of training."

The first resident to select the primary care genomics AOC was Dr. Jiefu Yuan, who currently visits the Clinic once a month to see patients. Dr. Yuan was born in China, but grew up in the Lancaster area, completed his undergraduate work at the University of Michigan, and graduated from the medical school at Drexel University. Dr. Yuan explains why he chose the primary care genomics AOC, "because I have had a long time interest in genetics since medical school. To be able to work with the Clinic is truly a special opportunity because of how unique the care is here. There really is no other place on the planet that provides the fantastic care and collaboration that the Clinic has."

Dr. Grace Loudon was the next LGH Family Medicine resident to select her AOC at the Clinic. Dr. Loudon grew up in the Poconos, received her undergraduate degree from Bryn Mawr College and graduated from The Sidney Kimmel Medicine College at Thomas Jefferson University. Dr. Loudon is in her second year at LGH, and is greatly enjoying her time at the Clinic. Dr. Loudon explains why she chose the Clinic, "When I was three and a half years old, my brother was born with Down Syndrome. Over the last 24 years, I have not only been his sister, but also his biggest advocate and supporter. I had always dreamed of being able to help patients with special needs, and the Clinic's AOC allows me to get that experience first hand. I am ecstatic to have this opportunity!"

Dr. Loudon hopes to have a career in taking care of complex patients with special needs. She enjoys that the Clinic allows her to be both a teacher and a learner. As she says, "You never know who will walk through the door and what challenges they might bring!"

When asked how residents benefit from AOC's, Dr. Ratcliffe explains, "Our residents acquire extensive knowledge about the conditions that are treated at the Clinic across the age spectrum. They receive exceptional experience in the recognition, diagnosis, and treatment of many genetically-based conditions. With their extensive experience in the care of adults, they provide needed general care for the adults receiving care at the Clinic."

Many of the "children" served at the Clinic over the last 29 years are now adults! Having doctors trained in adult medicine regularly seeing patients at the Clinic will enhance the Clinic's ability to provide appropriate and comprehensive care to patients as they age.

We'd like to thank Drs. Ratcliffe, Yuan and Loudon for their commitment to the Clinic and for their support!



Our Staff

Keturah Beiler, RN
Nurse

Lauren Bowser
Research Fellow

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

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

Julia Martin
Development Assistant

Erik G. Puffenberger, PhD
Laboratory Director

Teresa Rineer
Development Director

Donna L. Robinson, CRNP
Nurse Practitioner

Ashlin Rodrigues
Laboratory Technician

Emily Seitz
Scientific Grant Writer

Kevin A. Strauss, MD
Medical Director

Millie Young, RNC
Research Nurse

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Jacob Zook
Vice Chairman



Donor Spotlight

The Strasburg Rail Road

Let's travel back in time to the early 1800s in Lancaster County – the Philadelphia and Columbia Rail Road was one of the earliest in the area, and unfortunately bypassed the town of Strasburg by a distance of four to five miles. The townspeople of Strasburg petitioned to have a short line railroad connect to the Philadelphia-Columbia Rail Road and in 1832, the Strasburg Rail Road was chartered by PA legislature.

From the beginning of the Strasburg Rail Road until 1958, there was an ebb and flow of good times and bad. Following WWII, the construction of interstate and major highways decreased the use of railroads for travel and left the Strasburg Rail Road eventually in a state of near abandonment.

Upon hearing of the dismal fate of the Strasburg Rail Road, a group of 24 “rail fans” in Lancaster County bought the property and rail line. On a whim, they decided to buy an old wooden coach, remodeled it with a tiny gasoline engine, and started to take small groups of people on train rides through the Lancaster countryside. It was a successful combination of history, nostalgia, and Lancaster County countryside that garnered interest from the public. This winter their railyard should put the finishing touches on their 20th passenger car.

Several years ago, the Strasburg Rail Road chose the Clinic for Special Children as their “charity of choice” to donate funds from their Great Train Robbery and Christmas events. Steve Barrell, Stationmaster at the Strasburg Rail Road explained, “We are glad to support and partner with an organization right here in Lancaster County that has a local mission and works with children and families.”

The Strasburg Rail Road will feature its Great Train Robbery event on October 20 and all donations given during the event will benefit the Clinic. Another popular attraction, the Night Before Christmas trains, will be running on Thursdays and Fridays between November 29-December 21. A portion (\$5) of each adult ticket purchased for the Night Before Christmas trains benefits the Clinic. You can purchase tickets for these events in person at the Strasburg Rail Road or on their website (www.StrasburgRailRoad.com).

Since the initial partnership, the Strasburg Rail Road has **donated over \$36,000** to the Clinic for Special Children from the Great Train Robbery and Night Before Christmas trains and has made thousands of visitors to Lancaster County aware of the Clinic's mission and history. We are so thankful for the partnership with the Strasburg Rail Road and are excited to partner on future events together!

What's going on at the Clinic?



Thank you to over 280 runners along with sponsors and volunteers who raised over \$24,000 through our first 5k!



August was SMA Awareness Month! The Clinic staff wore purple to show our commitment to the research and treatment of Spinal Muscular Atrophy.



Dr. Kevin A. Strauss attended the GM3 family day in Indiana.



Over 100 Willow Valley residents attended a panel talk focused on MSUD from clinical, research, and patient perspectives.



A SMA family day featured talks from our doctors, researchers, collaborators and members of the pharmaceutical industry.



Members of the Clinic team attended the 2018 Translational Medicine in Plain Populations Conference in Madison, WI!

A note from our Community Liason, Lavina King.

A note from Lavina

Fall greetings! It's a beautiful time of year, I hope you are doing well with whatever journey you are traveling on . . .

I've now been working at the Clinic for two years and I've made lots of new friends that have enriched my life so much. I treasure those friendships and feel very blessed that God gave me the privilege to work for the Clinic for Special Children.

It's a learning process here at the Clinic. I've learned alot the past two years --- and hope to continue learning.

I'm here every Tuesday and Thursday. If there's (ever) anything I can do for you and your special Children, please let me know.

"With love and God bless you".



ClinicforSpecialChildren.org

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The Clinic for Special Children is a Section 501(c)(3) Public Charity for US Federal and State Tax purposes (Tax ID #23-2555373). Donations to the Clinic are tax deductible. Donors should consult their tax advisor for questions regarding deductibility. A copy of the Clinic's registration and financial information may be obtained from the PA Department of State by calling toll free, 1-800-732-0999.

2019 Benefit Auctions for the Clinic for Special Children

June 1 - Union County, PA

June 15 - Lancaster County, PA

June 22 - Shippensburg, PA

July 13 - Shiloh, OH

August 17 - Memphis, MO

September 14 - Blair County, PA

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 Wankroff Dr
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