



## Using Institutional Knowledge to Quickly Diagnose *TJP2* The Stoltzfus Family

“When Amanda was born we had no idea that she would be special or different,” Ada Ruth Stoltzfus explains of her daughter, Amanda. At around four months of age, Ada Ruth noticed that Amanda began to wake up frequently during the night and her quality of sleep declined drastically.

In addition to sleep issues, Amanda started to itch and scratch herself constantly. Ada Ruth tried cutting her nails, changing laundry and bath soaps, and anything else she could think of to alleviate Amanda’s itching. “As soon as we would try to bathe her, she would start itching and wouldn’t be interested in anything other than scratching her belly,” Ada Ruth explains.

When Amanda was six months of age, Ada Ruth heard of another family’s baby that had similar symptoms to Amanda. This family mentioned the possibility of vitamin K deficiency and recommended the Clinic for Special Children (CSC) to Ada Ruth, who then scheduled Amanda for her first visit shortly thereafter.

During Amanda’s first visit, Dr. Vincent Carson gave Amanda a vitamin K shot and ordered a targeted genetic test from CSC’s in-house laboratory to check for a condition called familial hypercholanemia (also sometimes called vitamin K deficiency) which can be caused by a variant in the *TJP2* gene. The condition can cause symptoms like itching, jaundice, poor weight gain, and bruising. This condition is known to

affect the Amish community so the CSC team knew the right test to order. Within hours, Amanda’s genetic test came back positive for *TJP2*.

*TJP2* is a disorder of the liver in which bile ducts are leaky and bile salts enter the bloodstream. When bile salts get into the bloodstream, it can cause extreme itchiness. In addition, fats from a person’s diet don’t absorb properly in the gut and can cause low levels of vitamins A, D, E, and K. When vitamin K is low, dangerous bleeding can occur.

Amanda was immediately placed on ursodiol to help her liver congestion and a vitamin called AquADEKs® to replace the vitamins her body could not absorb. This medication resulted in slight improvements to her liver inflammation and itchiness. At Amanda’s next visit with Dr. Laura Poskitt, she was also placed on a medication called rifampin.

Within a week after adding rifampin, Ada Ruth noticed a big difference in her daughter. “She was a lot happier, slept a lot better, and her scratching completely stopped,” Ada Ruth exclaimed. Today, Amanda is a happy baby who’s now teething, crawling, and pulling herself up. “We’re thankful that we were able to get an answer quickly with the Clinic,” says Ada Ruth.

Thank you to the Stoltzfus family for sharing Amanda’s inspiring story with *TJP2*!



## SPECIAL DATES

### Community Benefit Dinner Wednesday, October 14

Martindale Fellowship Center  
A seafood and ham take-out style dinner to benefit the Clinic! Details inside.

### 2020 Extraordinary Give Friday, November 20

ExtraGive.org  
Online Fundraiser - All Day  
Donate to the Clinic during this 24-hour giving day! Share about the Clinic with family and friends and set your own fundraising goal! Details inside.

### Thanksgiving - Office Closed November 26 & 27

All Day

### Christmas Eve - Office Closed Thursday, December 24

1 p.m. - 5 p.m.

### Christmas Holiday - Office Closed December 25 & 28

All Day

### New Years Eve - Office Closed Thursday, December 31

1 p.m. - 5 p.m.

### New Years Day - Office Closed Friday, January 1

All Day

As the situation around COVID-19 evolves, the event dates mentioned above could be subject to postponement or cancellation.

For any specific event updates, please call our office at 717-687-9407, check our social media accounts, or visit our website at [ClinicforSpecialChildren.org](http://ClinicforSpecialChildren.org).

## Staff News

### Vincent Carson, MD



Congratulations to Dr. Vincent Carson who was recently named the first Clinical Operations Director at the Clinic! In his new role, Dr. Carson will be joining the Clinic's Leadership Team and will be responsible for the day-to-day clinical operations, logistics, and services at the Clinic.

Dr. Carson joined the Clinic in 2016 as a Pediatric Neurologist. He grew up in Lancaster County and followed the work of the Clinic for years before joining our team.

Congratulations, Dr. Carson!

### Candace Kendig



Congratulations to Candace Kendig who was recently named the Clinic's Office Manager! In this role, Candace will coordinate the Clinic's front office operations. Candace joined the Clinic staff in 2017 as a Medical Receptionist and is currently pursuing a bachelor's degree in Health care Management at Central Penn College.

Congratulations, Candace!

### Janeen L. Maxwell, MPH



Welcome, Janeen! We're excited to welcome Janeen L. Maxwell, MPH to the Clinic team as our new Development Operations and Grants Strategist!

In this role, Janeen will be focusing on donor and institutional data management and grant development and administration.

Janeen has more than ten years' experience in grant writing, grant management, and non-profit capacity building. She holds a Master's of Public Health from West Chester University and a bachelor's degree in Psychology from Millersville University.

Welcome to the Clinic, Janeen!

## CSC Community Benefit Dinner

**Wednesday, October 14**

**4:00 p.m. – 7:30 p.m.**

**Take-out only**

**at the Martindale Fellowship Center**

We are looking forward to our 2020 Community Benefit Dinner on Wednesday, October 14<sup>th</sup> from 4:00 p.m. until 7:30 p.m. at the Martindale Fellowship Center (352 Martindale Rd., Ephrata, PA 17522)! Due to COVID-19 precautions, the dinner will be take-out only this year. All proceeds will benefit the Clinic.

Take-out meals will include a variety of delicious food like oysters, shrimp, ham, cole slaw, green beans, fresh fruit cup, rolls, and a whoopie pie! The cost of the dinners is by donation only.

RSVP's are recommended but not required. To RSVP, please call the Clinic at 717-687-9407 or email [RSVP@clinicforspecialchildren.org](mailto:RSVP@clinicforspecialchildren.org).

### **It's Flu Shot Season**

It's that time of the year! Anyone over six months of age is encouraged to get an annual flu shot. We offer shots to our patients and their immediate family members for \$20 per person.

The best way to protect yourself and your family is to avoid exposure, practice good hand-washing, and get your annual flu shot.

Please call the Clinic at 717-687-9407 to schedule your flu shot!

### **WiTNness Natural History Study Amish Nemaline Myopathy (ANM)**

One of the genetic conditions that we see all too commonly at the Clinic for Special Children is Amish Nemaline Myopathy (ANM), commonly known as "chicken breast disease." This genetic disorder affects about 1 in every 500 babies in the Amish community.

It is caused by a genetic error in the *TNNT1* gene responsible for making troponin, a protein important in muscle contraction. Children with ANM are unable to make this protein, which causes progressive muscle weakness and causes them to die early in childhood.

The serious nature of this condition and the number of Amish babies it affects have made

the disorder a priority for the Clinic to work with other experts to develop a life-changing therapy. As part of that process, we need to better understand the different ways that ANM affects the health and wellbeing of children with the disorder. To accomplish this, we launched the WiTNness study. The study is designed to capture how this condition affects growth and development over time. There are currently no medications that can cure ANM so participating infants do not receive any medicine as a part of this study, however careful data collection is a critically important foundation for the development of meaningful therapies.

### **Designing a Gene Therapy for ANM**

In addition to this work with ANM patients, we are also working with other medical and scientific collaborators to understand how the lack of the troponin protein in ANM causes muscle weakness and to identify possible therapies specific for this condition. Currently, we are working with researchers at the University of Massachusetts Medical School and Tufts Veterinary School to develop a gene therapy for ANM.

Gene therapy is a new way to treat, and potentially cure, genetic disorders. Gene therapy uses a modified, harmless virus to insert a working copy of a gene into target cells, allowing the functional protein that was lacking to be produced and correcting the genetic error at the root of the condition. For ANM, gene therapy would introduce a working copy of the *TNNT1* gene. The working gene would direct the cells to produce the troponin necessary for muscle contraction. Our hope is that this kind of treatment would significantly improve the lives of children with ANM.

Lastly, we are also in touch with other doctors around the world who treat non-Amish patients with ANM to learn about their experiences. Our hope is that we can all work together to make a real difference in the lives of the precious children with ANM. Please contact the Clinic for Special Children at 717-687-9407 and ask for Mariah if you have questions or want to learn more about our work in ANM.





**Dr. Grace Loudon**  
*A Family Practice Physician joins the CSC team!*

We are excited to announce that Grace Loudon, MD, joined our staff full-time in August. Dr. Loudon, a trained family medicine physician, is an important addition to our staff. Many of the children we've cared for at the Clinic over the years are now adults with complex medical needs. The addition of a doctor with expertise in family medicine at the Clinic allows our pediatric providers to collaborate with an expert in adult care in order to provide lifelong, comprehensive care for our existing patients. It also allows the Clinic to offer care to new adult patients with complex genetic disorders.

*“The Clinic is patient care, research, and science at its finest. I am ecstatic to be here working with, learning from, and being inspired by my patients, their families, and my colleagues.”*

Dr. Loudon explains her passion for the Clinic, “The clinic is patient care, research, and science at its finest. I am ecstatic to be here working with, learning from, and being inspired by my patients, their families, and my colleagues. But actually, it’s not work because I love what I get to do.”

Dr. Loudon’s training allows her to see patients of any age making her an ideal addition to our staff. Her experience in working with adult patients will allow her to lead efforts in formalizing an adult services program at the Clinic. Dr. Loudon earned a bachelor’s degree in biology from Bryn Mawr College in 2012 and a medical degree from Sidney Kimmel Medical College at Thomas Jefferson University in 2017. She completed her family medicine residency at Penn Medicine Lancaster General Health (LGH).

Dr. Loudon is already a familiar face at the Clinic, as she has been visiting and seeing patients periodically since mid-2018 as part of a special program between the Clinic and LGH called the Clinical Genomics area of concentration.

In her free time, Dr. Loudon enjoys spending time with her family and her two cats, visiting the beach, working on home improvement projects, biking, and running with her dog, Remi. All of us at the Clinic are excited to welcome Dr. Loudon to our team!

2020  
**Extraordinary Give**

**THE EXTRA<sup>TM</sup>  
ORDINARY  
GIVE**



**Friday, November 20<sup>th</sup>**  
Midnight - 11:59 PM  
ExtraGive.org

We invite you to donate to the Clinic during Lancaster County’s largest day of online giving! The ExtraGive benefits more than 500 local non-profit organizations and every dollar donated through ExtraGive.org on Friday, November 20<sup>th</sup> will be stretched by a pool of \$500,000 from local sponsors. We also have our own matching dollars for Clinic donors thanks to Nemours/A.I. duPont Hospital for Children and Earl R. Martin, Inc.!

Donating is simple. On Friday, November 20<sup>th</sup> visit ExtraGive.org and select ‘Clinic for Special Children’. If you would prefer to donate over the phone, please call us on Friday, November 20<sup>th</sup> between the hours of 9 a.m. - 5 p.m. and we would be happy to receive your gift over the phone. Won’t have access to internet on the day of the ExtraGive? Call us before or on November 20<sup>th</sup>, and we’ll process your ExtraGive donation for you on the day of ExtraGive.

More details on how to participate in the ExtraGive are included with the insert in this newsletter. Thank you for your support!







2019 MSUD Family Day Group Photo

## Groundbreaking 30-Year MSUD Study

*Recently Published Research from the Clinic*

For more than 30 years, the Clinic for Special Children has been an international leader in the treatment and research of maple syrup urine disease (MSUD). MSUD is a rare genetic disorder that interferes with the body's ability to process protein and can result in irreversible brain damage. Throughout the Clinic's history, doctors and researchers have made substantial improvements in the treatment and care of this disorder.

Earlier this year, the Clinic led a broad collaborative effort to publish the largest study of MSUD to date, which summarizes three decades of MSUD research and clinical care. The research was published in the scientific journal, *Molecular Genetics and Metabolism*, and summarized data for individuals followed over 30 years. The data reported in the study includes survival rates, hospitalization rates, metabolic crises, liver transplantation, and cognitive outcome.

Large studies, like the Clinic's MSUD study, that collect historical data are important because they provide an understanding of the natural course of a disorder based on the current standard of care. When new therapies are created, this research can be used as a "control group" in which to judge the effectiveness of a new treatment. Should potential treatments for MSUD be developed in the future, this study will provide researchers from around the world with vital benchmark information.

Before the Clinic's founding, one in three children born with MSUD died from neurological complications before the age of 10 years old and the majority of survivors were permanently disabled. The three decades of MSUD clinical care

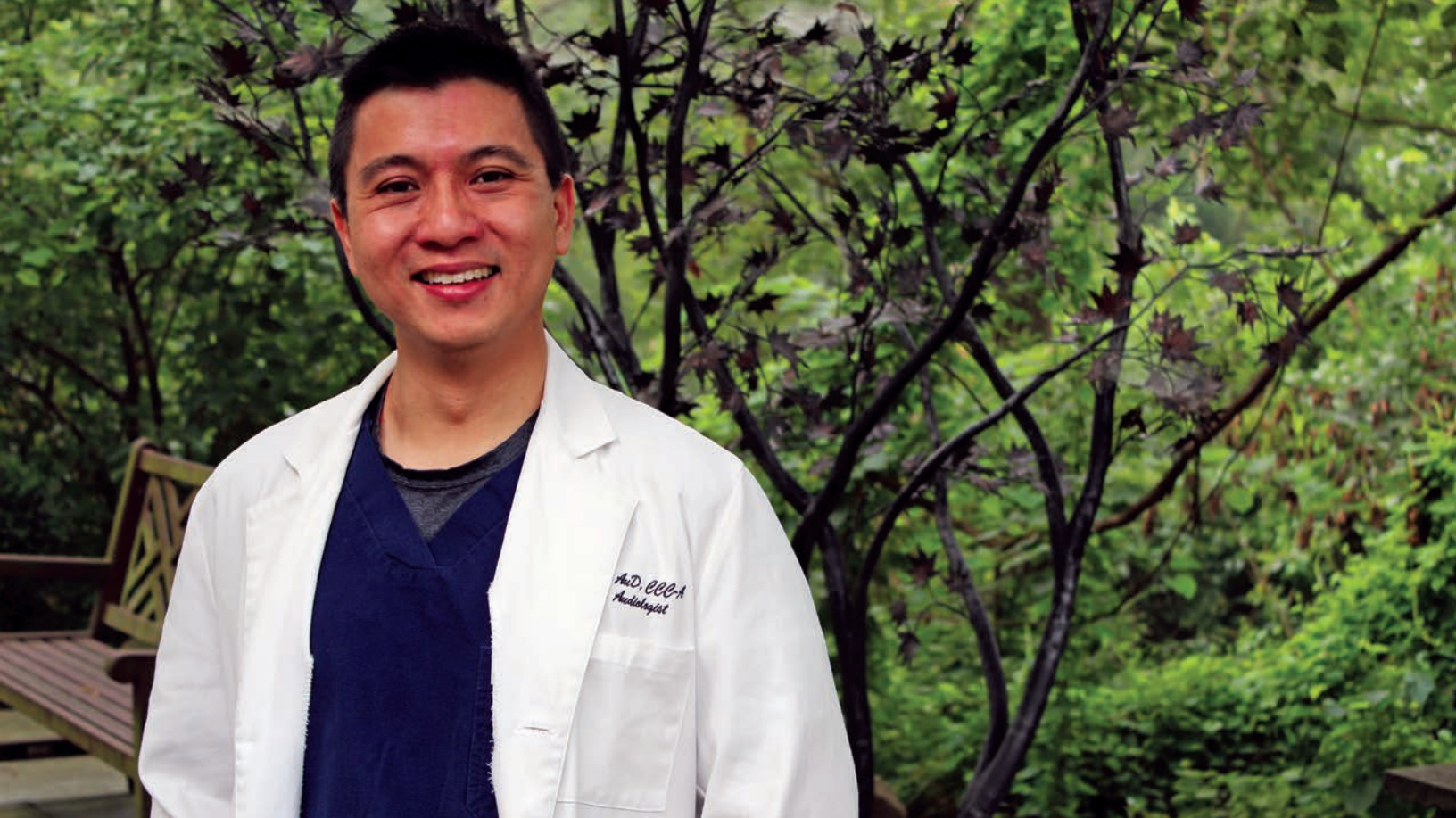
and innovation by the Clinic has increased survival rates from 63% to 95% while hospitalization rates have decreased from 7 to just 0.25 hospital days per patient per year.

Innovations in MSUD treatment at the Clinic have included new prescription formulas for children and adults as well as advancements in elective liver transplantation – a collaboration with the Hillman Center for Pediatric Transplantation (UPMC Children's Hospital of Pittsburgh). A total of 93 MSUD patients have been transplanted since the start of this collaboration in 2003 with an overall graft and patient survival rate of 100%.

The study analyzed more than 13,859 amino acid profiles, a blood test that MSUD patients use to determine the levels of protein in their body. Similar to a diabetic glucose test, these amino acid levels provide results to inform of dietary changes needed to stabilize protein levels. Too much protein can cause a toxic build up of chemicals in the brain that cause permanent brain damage. The study found that while strict dietary limitations and the use of prescription formula keep levels in acceptable ranges, it is hard to sustain these changes lifelong.

Overall, while there have been life-changing innovations in the treatment and care of MSUD, it still remains a morbid and potentially fatal disorder. There remains a critical unmet need for a safer and effective disease-modifying therapy, like gene replacement or editing therapies. At the Clinic, we are working each day to advance the treatment of disorders, like MSUD, to provide a better quality of life for our patients.





### **Collaborator Spotlight:** Hui Shing Andy Lau, AuD, CCC-A, Audiology, Nemours Children's Health System

A personality profiling test in college led Dr. Hui Shing Andy Lau to discover his love for the field of audiology. Dr. Lau, a graduate of The College of William and Mary in Virginia, worked for a few years after college as an Audiology Assistant before deciding to obtain his doctorate in audiology. He completed his doctoral degree in audiology at Salus University in Elkins Park, PA.

Before settling in the USA for his education, Dr. Lau grew up in Canada and Hong Kong. He describes himself as “a bit of a nomad” as he has moved about every four years throughout his time in the USA.

Dr. Lau is currently a Pediatric Audiologist at Nemours/Alfred I. duPont Hospital for Children in Wilmington, DE. He first learned of the Clinic for Special Children (CSC) from his colleague, Thierry Morlet, PhD, a Senior Research Scientist and Head of the Auditory Physiology and Psychoacoustics Laboratory at Nemours.

When Dr. Lau first joined the team at Nemours, he jumped at the opportunity to visit CSC as he “had always been curious with how to make audiology accessible akin to the way rural doctors do with medicine, and visiting CSC was a golden opportunity for me.” Dr. Lau currently visits CSC one day a month with a team from Nemours including Dr. William Parkes, an Otolaryngologist.

A typical day at CSC for Dr. Lau includes performing hearing evaluations on patients; prescribing, fitting, and checking hearing aids; and conducting newborn hearing

screenings. Dr. Lau normally works most of the day in CSC's audiology booth. The most common diagnoses Dr. Lau sees at CSC are hearing loss secondary to a history of reoccurring ear infections and hearing loss associated with syndromes. Since the audiology team is at CSC once a month, their schedules are usually booked full and the day goes by very quickly.

When asked why he's passionate about CSC, Dr. Lau explains, “I love the Clinic for the fact that it is pure and wholesome. It is an opportunity to practice my chosen profession in the most organic way. When I am at the Clinic, I get to enjoy the interactions with my patients and the rapport that we build along the way. I get to witness and partake in providing compassionate care that uplifts their quality of life.”

Dr. Lau explains how he feels after he works at CSC, “For me, I leave the Clinic each time feeling fulfilled, feeling I have done my part in being the good that this world needs. In return I have been embraced by the families and garnered respect from my peers. It is a unique experience, and in my heart, the Clinic is a magical place made special by those who work there and the community that entrust its welfare to it.”

In his free time, Dr. Lau enjoys hiking, taking pictures, or doodling. We are so grateful to Dr. Lau for providing audiology care to the patients of CSC!



## Our Staff

Keturah Beiler, RN  
Nurse

Karlla Brigatti, MS, CGC  
Research Operations Director

Vincent Carson, MD  
Clinical Operations Director

Kelly Cullen  
Communications Manager

Mariah Everett  
Research Associate

Adam D. Heaps, MS, MBA  
Executive Director

Christine Hendrickson, RNC  
Nurse

Candace Kendig  
Office Manager

Lavina King  
Community Liaison

KaLynn Loeven  
Laboratory Scientist

Grace Loudon, MD  
Family Practice Physician

Julia Martin  
Development Associate

Janeen L. Maxwell, MPH  
Development Operations and Grants Strategist

Laura Poskitt, DO  
Pediatrician

Erik G. Puffenberger, PhD  
Laboratory Director

Stephen D. Ratcliffe, MD, MSPH  
Senior Consulting Physician

Donna L. Robinson, CRNP  
Nurse Practitioner

Ashlin Rodrigues  
Laboratory Scientist

Caitlin Russell, MS, MPH, CGC  
Genetic Counselor

Emily Seitz  
Development Director

Kevin A. Strauss, MD  
Medical Director

William Van Ess, MS, CFE  
Accounting Manager

Dawn Wade  
Medical Receptionist

Millie Young, RNC  
Research Nurse

## Board of Directors

Cindy Bo, MBA  
Chair-Charity Committee and Secretary

Herman Bontrager  
Chairman

Peter Crino, MD, PhD

Leon Hoover  
Vice Chairman

Leonard Hurst

Mark Martin  
Treasurer

Jacob Petersheim

Stephen Tift, MD

Glen Zimmerman  
Chair-Development Committee

Jacob Zook



## Donor Spotlight

### Weaverland Youth for Truth Committee

For over 20 years, the Weaverland Youth for Truth committee (WYT) has been instrumental in supporting the Clinic for Special Children and other local non-profits in the Lancaster County area. The committee is comprised of 10 youth members, ranging in age from 17–25 years. Each year the committee is elected to serve and focuses on two main areas: helping with local food stands at benefit events and general service projects in the community.

Each year the WYT committee supports eight benefit sales by managing food stands for the auctions. At the Clinic's Lancaster County benefit auction each year, the WYT committee runs the french fry, soup, and other food stands to help raise money for the Clinic. The committee has also pitched in with organizing the french fry stand at the Clinic's Fall Family Fun Day in past years.

In addition to lending a helping hand at community events, the committee manages its own three major fundraising events each year, with proceeds benefiting local nonprofits. These fundraisers include a ham and cheese sale, a spelling bee, and a pizza fundraiser. The committee generally sells over 10,000 sandwiches and 3,000 pizzas – an impressive feat! The spelling bee takes place each year at the Martindale Fellowship Center and any school-aged children can sign up to participate. A supper is also held during the evening with over 100 attendees. The proceeds from the event are distributed to various local charities through the WYT committee.

During the COVID-19 pandemic this year, the committee has been responsive to needs in the community. In May, they organized and distributed food boxes to those in need in Lancaster City. Even though many of the fundraising events that the committee participates in were cancelled, they have risen to meet the challenge. Within two weeks in June, the committee organized an outdoor chicken BBQ and car wash event to support the Clinic as our 2020 Lancaster County benefit auction was cancelled. They grilled over 1,500 chicken legs, sold 1,000 meals, and washed over 100 cars during the event!

The committee's event raised thousands of dollars for the Clinic during an uncertain year and we are so thankful to the committee for their steadfast support. We are inspired by this group of youth who are positively impacting their community!



# 2020 VIRTUAL 5K

Thank you to our participants & sponsors for raising over \$10,000!

*Enjoy the below photos that were submitted by virtual 5k participants!*



## THANK YOU to our 2020 Virtual 5k sponsors!

### Gold Sponsors



### Silver Sponsors



### Bronze Sponsor



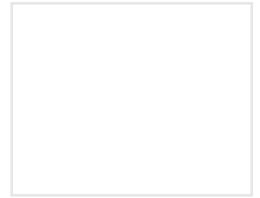


# Clinic for Special Children

535 Bunker Hill Road  
PO Box 128  
Strasburg, PA 17579

tel (717) 687-9407  
fax (717) 687-9237

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



The Clinic for Special Children is a Pennsylvania non-profit corporation and a 501(c)3 public charity for US federal and state tax purposes (Tax ID # 23-2555373). The official registration and financial information of The Clinic for Special Children, Inc. may be obtained from the Pennsylvania Department of State by calling toll free, within Pennsylvania, 1 (800) 732-0999. Registration does not imply endorsement.

## THANK YOU!

This year has been a challenging year for us all - one that has been uncertain and unpredictable. For us at the Clinic, a defining moment gave us hope and positivity and it was because of donors, like you.

In April, we had to start cancelling fundraising events and benefit auctions, which account for over 60% of our annual philanthropic budget. After deciding to cancel events to protect our patients, staff, and supporters, we sent an appeal to friends of the Clinic.

The response was overwhelming! With your support, we are at 97% of our pre-COVID auction goal at the end of September. Your support allowed us to continue to serve our vulnerable patients during a time when they needed us the most. Thank you for your unwavering support of children and adults with rare genetic disorders.



### 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."

