Clinic for Special Children®

Newsletter

ISSUE #65

SUMMER 2025

Pictured: Cassandra Martin

Long-awaited Answers
The Martin Family

When Carolyn Martin first brought her 25-year-old daughter, Cassandra, to the Clinic for Special Children, she left the appointment feeling amazed. "The doctors, nurses, and genetic counselor asked all sorts of questions about Cassandra. They were interested in every part of her health, and I could tell they cared about her. They shared my concerns and I felt very listened to," shares Carolyn.

The Martin family's path to finding the Clinic included decades of specialist appointments, tests, and uncertainty. "Pretty early on in Cassandra's life, we noticed she had developmental delays. As the years progressed, we dealt with eating challenges, anxiety, seizures, disturbing behaviors, eye issues, and more. Cassandra would barely leave the house, and we were at a loss for how to help her. We experienced many years where we had more questions than answers," explained Carolyn.

After pursuing many different healthcare options for Cassandra, Carolyn's friend shared her positive experiences with the Clinic. With hope, Carolyn called the Clinic to have Cassandra evaluated. "I didn't understand how much the Clinic could offer Cassandra until Dr. Grace Meier explained the different ways in which we could work together. Since we started at the Clinic, Dr. Meier has grown into being Cassandra's primary care provider, her geneticist, providing her OB/GYN and neurology care, navigating us through guardianship, connecting us with specialists, and so much more," shares Carolyn. "The Clinic is just a gift to us; you are my family's people."

To try to find a genetic answer for Cassandra, Amy Albright, a genetic counselor at the Clinic, walked the family through our pipeline of progressively in-depth genetic testing. After conducting a test that looked at all approximately 20,000 genes that code for proteins in the body, the Clinic team found a genetic change of interest in a gene called *GPATCH11*. There wasn't much information about *GPATCH11* and its impact on health. Fortunately, around the same time, the first scientific paper had been published describing a group of several people with similar *GPATCH11* changes as Cassandra. Amy called Carolyn to share the findings of the paper and how it related to Cassandra's results. They agreed that Cassandra and the individuals from the paper shared striking similarities.

In a surprising turn of events, Carolyn met another family whose child was recently diagnosed with *GPATCH11* at Wills Eye Hospital (WEH) in Philadelphia, and due to the rarity of *GPATCH11*, the genetic counselor at WEH contacted Carolyn to get more information about Cassandra. Now, the WEH team and Clinic staff are collaborating on research efforts to share more about *GPATCH11* and its effects on health. The Clinic laboratory has also added *GPATCH11* to its carrier test, the Plain Insight Panel™.

When asked how she's doing today, Cassandra shares, "I am joyful and not complaining as much. I like this version of me. I'm taking care of my silly worries when I have them."

SPECIAL DATES

Ohio Benefit Auction
Saturday, July 12
Blooming Grove Auction Inc. | Shiloh, OH

Missouri Benefit Auction
Saturday, August 16
Ed Good Family Farm | Memphis, MO

Labor Day - Office Closed
Monday, September 1
All Day

Blair County Benefit Auction
Saturday, September 13
Morrison's Cove Produce | Roaring Spring, PA

Finger Lakes Benefit Auction
Saturday, October 4
Ontario Produce Auction | Stanley, NY

Community Benefit Dinner
Tuesday, October 21
Martindale Fellowship Center | Ephrata, PA

The Extraordinary Give
Friday, November 21
All Day
www.ExtraGive.org

Thanksgiving - Office Closed
November 27 & 28
All Day

Christmas Eve - Office Closed
Wednesday, December 24
Office closes at 1:00 p.m.

Christmas - Office Closed
December 25 & 26
All Day

New Years Eve - Office Closed
Wednesday, December 31
Office closes at 1:00 p.m.

Events are subject to change due to severe weather conditions, restrictions, or unforeseen circumstances.

Recently Published Research Studies

Rare genetic variant tied to risk of Major Depressive Disorder (MDD)

Researchers from the Clinic, Columbia University, and Regeneron Genetics Center collaborated on a study recently published in the journal *Proceedings of the National Academy of Sciences* that identified a variant in the gene *GPR156* in a large Mennonite group that doubles the risk of major depression in those who carry it.

Major depressive disorder is a leading cause of disability worldwide and can affect people from all communities and backgrounds.

This research deepens our understanding of the causes of depression and introduces a new target for its treatment. GPR156 is expressed in a deep part of the brain known to play a crucial role in behavioral responses to stress.

Immunopathological and microbial signatures of inflammatory bowel disease in partial RAG deficiency

In early May, researchers from the Clinic, the National Institutes of Health (NIH), the University of South Florida, and other collaborators published research in the *Journal of Experimental Medicine* that describes the immune and microbiome characteristics of patients with inflammatory bowel disease (IBD) due to partial RAG deficiency caused by genetic variants in the *RAG1* or *RAG2* genes.

They found that the immune system becomes too aggressive and the helpful bacteria in the gut are missing or unbalanced.

Regular medicine did not help much, but bone marrow transplant was able to fix the immune and gut problems, showing it may be the best treatment for patients with genetic RAG deficiency and IBD.

For more information on these papers, please contact us at 717-687-9407 or visit the 'Published Papers' section on our website at www.ClinicforSpecialChildren.org.

Measles Reminder

As we continue to monitor the national measles outbreak and cases in Lancaster County, we wanted to share a couple of reminders.

We regularly offer vaccine clinics for anyone uninsured (you don't have to be a Clinic patient). The cost is $20 per person and the fee covers all vaccines needed in one visit. We especially encourage the measles vaccination (MMR vaccine) amid the current outbreak. To schedule, call us at 717-687-9407.

If you have an upcoming appointment at the Clinic and you or a family member are experiencing fever, red eyes, cough, and a full-body rash that starts on the head, please call our office at 717-687-9407 before coming in for your appointment because the measles virus is highly contagious.

Matching Gift Opportunity!

New Lab Equipment Needed

We are excited to share that we have a 1-for-1 giving match available up to $20,000, thanks to a local family foundation.

The match opportunity is specifically for helping us purchase a new laboratory machine, called an HPLC (high-performance liquid chromatography), that provides critical monitoring tests for patients with metabolic disorders, like maple syrup urine disease. The test allows us to adjust diets and treatments to prevent potentially deadly buildups of amino acid levels in the body.

One of our current machines, over 10 years old, is no longer being supported by the manufacturer. To keep up with the current demand for testing, we need $80,000 to purchase a replacement.

To learn more about this opportunity, please visit page 6 of this newsletter. To give towards the match, please note "New Lab Machine" in your check memo or your online gift. On the enclosed giving envelope, you can also check a box to designate your gift for this opportunity. Thank you for your support!

Remaining Family Days

We're looking forward to welcoming patient families and collaborators to the Clinic for our remaining family days of 2025! Each family day combines expert talks with a day of fellowship focusing on a specific rare genetic disorder.

Families with a member diagnosed with properdin deficiency, CFI deficiency, or myotonic dystrophy type 1 should receive invites in the mail. If you have any questions, please contact Kelly Woodson, Events Manager, at 717-687-9407 or kwoodson@clinicforspecialchildren.org.

Properdin & CFI Deficiency Family Day
Wednesday, July 16th

Myotonic Dystrophy Type 1 Family Day
Wednesday, August 13th

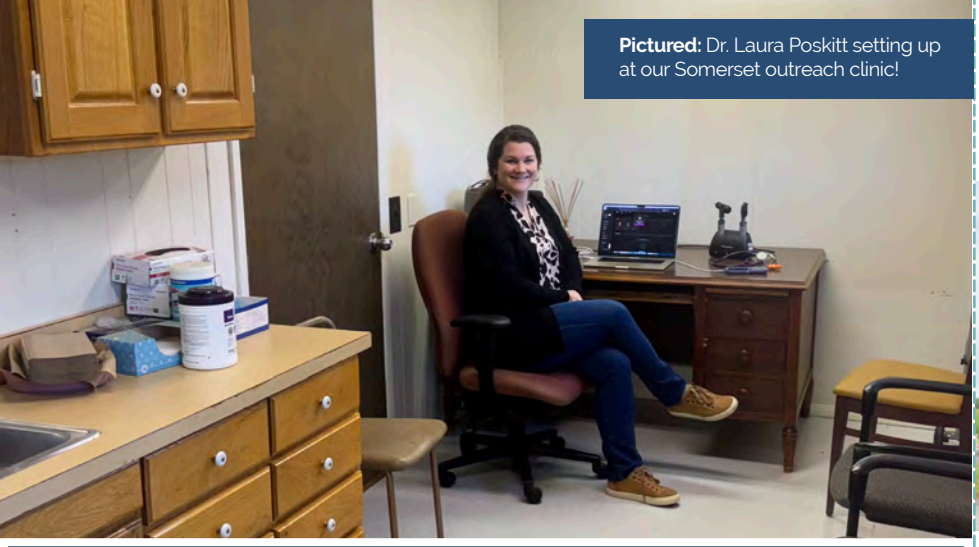
Amish Research Clinic

Looking for participants for a new wellness study

The Amish Research Clinic (ARC), a collaborator of the Clinic for Special Children, is looking for participants from the Plain community for a new wellness study, "Characterizing Genetic Diversity in the Plain Populations."

This study is organized by the ARC (not a study affiliated with the Clinic for Special Children). The goal is to understand how genes affect the risk of heart disease and other health conditions. Participating takes less than one hour and is a one-time visit. Participants will receive blood work results to give them an overview of their current health at no cost to them. You are eligible to participate if you are 18 years of age or older, of Plain Anabaptist ancestry, and have not participated in this study previously.

For more info or to schedule an appointment, please contact Charlene Wolford, BSN, RN at 717-682-0636 or the ARC at 717-392-4948.



Pictured: Dr. Laura Poskitt setting up at our Somerset outreach clinic!

Expanding our Outreach

Providing care closer to home

Central to the mission of the Clinic is providing affordable and accessible care. Our outreach program expands our reach to meet patients where they are - increasing accessibility to our services. We're excited to share several new updates about our outreach program! Christine Hendrickson, RN, BSN, PED-BC, continues in her role as a nurse at the Clinic, but now has an extra focus on managing our outreach clinic program. We're also excited to announce that we have launched new outreach clinic sites this year in Memphis, MO, Penn Yan, NY, and Newville, PA!

The outreach clinics scheduled for the rest of this year are shown below. A few important notes regarding our new outreach clinics for this year: you must have already been seen as an established patient at our Clinic in Gordonville. Spots are limited, so you must have a scheduled appointment to be seen at an outreach clinic (no walk-ins). If you are an eligible patient family residing near one of our new outreach clinics, we will be contacting you with details. If you have any questions regarding outreach clinics, please call us at the Clinic at 717-687-9407.

2025 Dates

- NEW -

Memphis, MO *(Dr. Vincent Carson)*

August 15th

- NEW -

Penn Yan, NY *(Dr. Grace Meier)*

October 3rd

Mifflinburg, PA *(Drs. Vincent Carson & Julia Goroff)*

Monthly on 3rd Thursdays

July 17th, August 21st, September 25th, October 16th, November 20th, & December 18th

- NEW -

Newville, PA *(Dr. Grace Meier)*

November 11th

Somerset, PA *(Dr. Laura Poskitt)*

August 11th & 12th, November 3rd & 4th

Remaining 2025 Benefit Auctions

Most auctions will feature CSC remarks and quilts midday.

Events are subject to change due to severe weather conditions, restrictions, or unforeseen circumstances.

Saturday, July 12
Ohio Auction

Breakfast at 7:00 a.m., auction at 9:00 a.m.

Blooming Grove Auction Inc.
1091 Free Road, Shiloh, OH 44878
Contact: Michael Newswanger | 419-896-2184

Saturday, August 16
Missouri Auction

Airplane Candy Drop at 9:30 a.m.
Auction at 10:15 a.m.

Friday Evening Event
Aug. 15 | Food (5:00 p.m.) + Children's Auction (7:00 p.m.)

Ed Good Family Farm
10507 County Road 813, Memphis, MO 63555
Contact: Harlan Burkholder | 660-341-4113

Saturday, September 13
Blair County Auction

Breakfast at 7:00 a.m., auction at 8:30 a.m.

Friday Evening Event
Sept. 12 | Starts at 4:30 p.m.
Rib Dinner & Auction Preview

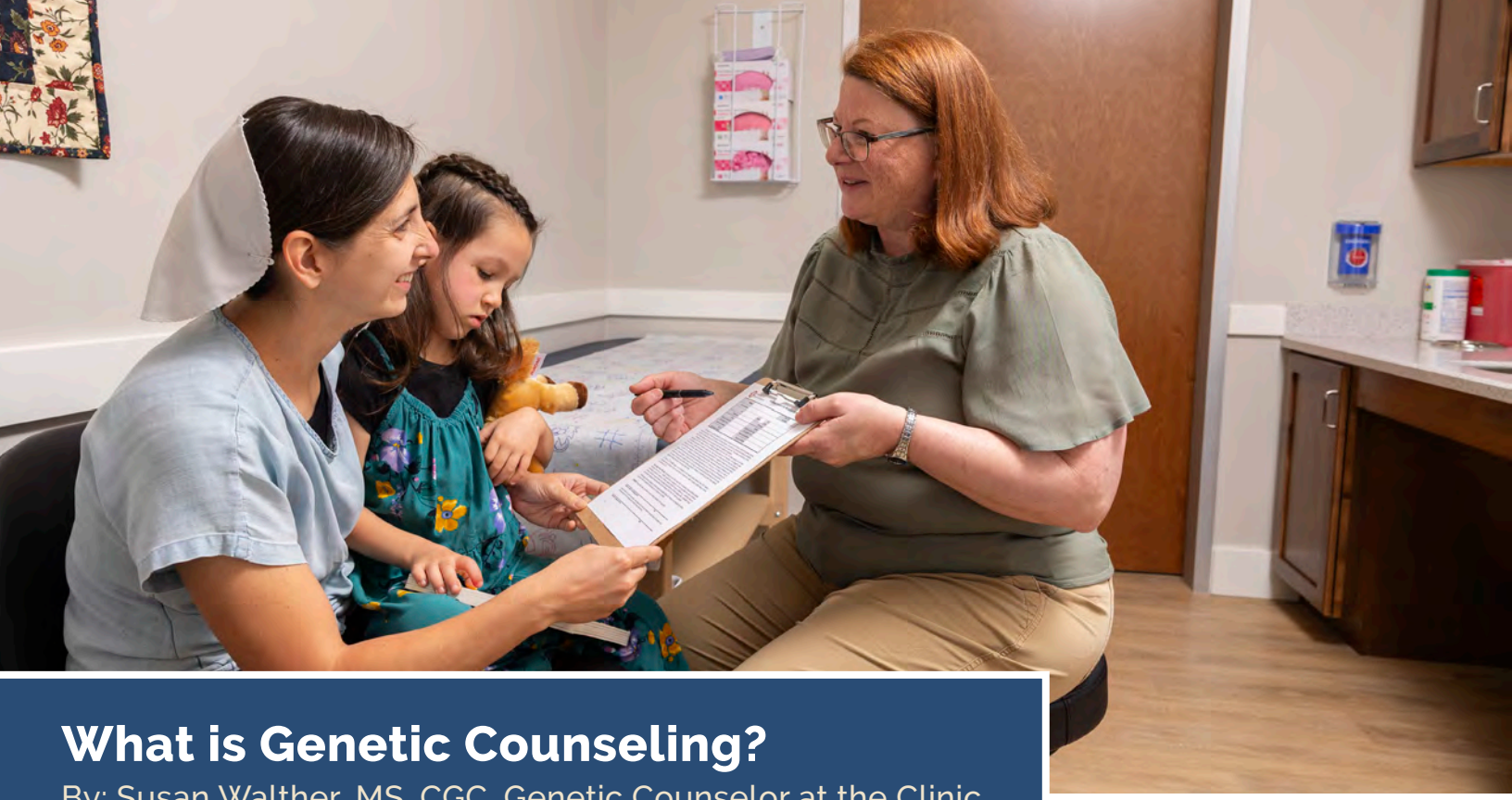
Morrison's Cove Produce
4826 Woodbury Pike, Roaring Spring, PA 16673
Contact: Mervin Martin | 814-793-3529

Saturday, October 4
Finger Lakes Auction

Country breakfast at 6:30 a.m., auction at 8:00 a.m.

Friday Evening Event
Oct. 3 | Starts at 4:30 p.m.
Grocery & Craft Auction + Rib Dinner

Ontario Produce Auction
4860 Yautzy Road, Stanley, NY 14561
Contact: David Fox | 585-526-5913



What is Genetic Counseling?

By: Susan Walther, MS, CGC, Genetic Counselor at the Clinic

When a patient has their first visit here at the Clinic, the appointment usually runs about two hours long! In addition to meeting with a nurse and doctor, the family speaks with one of our genetic counselors. These specialized healthcare providers ask about family history, review testing options, and more! Learn more about genetic counseling and why it's a cornerstone of the care we provide here at the Clinic.

What is genetic counseling?

Genetic counseling helps give families an understanding of how genetics might affect their health or the health of their future children. The genetic counseling process involves collecting family medical histories, providing information about genetic testing options, sharing information about genetic conditions, and guiding individuals and families in making informed decisions about their health relating to genetics.

Understanding the basics of genetics

The human body has approximately 20,000 genes in each cell. Genes carry instructions for how people develop, grow, and maintain health. Our bodies read through each gene instruction to create and build proteins. Gene instructions provide information for how the proteins function and the amount of proteins needed to maintain health. These instructions are a series of letters representing building blocks (molecules), and the specific combination of letters determines which protein is made. Therefore, if letters are changed, removed, or added, the body's ability to read through the gene instruction is interrupted and can negatively affect the resulting protein.

Why is understanding differences in genetics important to health?

When gene instructions are changed or interrupted, the effects on health can be significant. When healthcare providers know about genetic differences in a family, genetic testing can be performed to determine which family members need specialized medical care. It can be critical to know about genetic differences before or soon after a baby is born because specialized medical care can minimize or prevent serious health issues and can improve overall quality of life.

For children and adults who come to the Clinic with an unknown cause for their health concerns, genetic testing can often help provide answers. Genetic counselors work with families and their medical providers to better understand genetic conditions with the goal of developing an appropriate plan for care. When a genetic cause for health issues is known, it allows medical providers to read medical reports on similar patients, which decreases the guesswork on managing symptoms and provides guidance on anticipated symptoms that might develop in the future.

Why is carrier screening important?

An important test designed and conducted through our Clinic's on-site laboratory is the Plain Insight Panel™ (PIP). This test is specific for genetic variations found in the Amish and Mennonite communities. It is ideal for young couples who are thinking of starting a family. PIP testing for couples can identify genetic matches that pose a risk for their children, and a plan can be discussed with the couple's midwife for testing babies at delivery through an umbilical cord blood sample. When we know exactly what genetic disorder(s) the baby is at risk for, we can quickly run a targeted test through our laboratory and provide parents answers within 24 hours or less.

Most couples who decide to do PIP testing find out they are carriers for several genetic conditions, but each carries for different disorders and they *do not* match. In these results, much of the known genetic risk in the Plain community is eliminated, and parents can be reassured about the health of their children.

Meet our Genetic Counselors

Susan Walther, MS, CGC & Amy Albright, MS, CGC

We sat down with our genetic counselors at the Clinic, Susan (S) and Amy (A), to learn more about their career journeys and the field of genetic counseling. The interview was edited for length and clarity.

Why did you choose genetic counseling as a career?

S: I had been in a laboratory research role at a large university when I found out about the field of genetic counseling. I had a desire to move into a new role where I could use my science knowledge to work directly with patients in a healthcare setting. Genetic counseling was a great career to combine those skills and knowledge bases.

A: In college, a family member told me about their positive experience with a genetic counselor during their pregnancy. When I looked at the field of genetic counseling, as Susan mentioned, I liked the blend of science and people skills. After shadowing genetic counselors, I knew this field was the right fit for me.

What do you like the most about your role?

S: The most interesting and challenging part of my job is getting a new test result that we haven't seen at the Clinic before. I research to understand the gene and its clinical presentation, and then think through how to explain it to the family. I then work with our physicians to develop the best care plan for the family.

A: Though it is also challenging, I appreciate being able to help families find answers. Oftentimes, parents will come in with concerns, and part of my job is validating their instincts and then helping find an underlying genetic cause for the differences they are noticing in their child. It is rewarding to help families navigate that process.

What are some of the biggest challenges in your role?

A: It's hard when we identify a genetic answer, but it does not have a cure, and it's life-limiting. It's also really challenging when a family has obvious health concerns about their child, but we are unable to find a diagnosis. In both of those situations, being there for families and helping them in whatever way we can is all we can do.

S: Even if the news that I have to deliver is unexpected, our role in being able to provide kindness and offer a medical home here at the Clinic helps with these challenges. In some cases, the potential for future research studies or treatments can help provide hope for us, and more importantly, for the family.

What is the most rewarding part of being a genetic counselor?

A: Being able to finally give an answer to a family that's been searching for years and has been on a long medical journey. Also, being able to call a family with negative cord blood results, especially if they previously had affected children.

S: I agree that it's being able to find an answer and reporting out negative cord blood results. I'm grateful when parents notice something may be happening with their child, like developmental delay, and they trust that they can come to us to tell that story and we can help them. We're listening and we will do something.

What do people commonly misunderstand about genetic counseling?

A: People may assume that genetic counselors want to sell or push genetic testing, but that is not our goal. We are here to present all your testing options and help you make a decision that aligns with your values and helps your healthcare team provide the best care for your family.

S: One of the tenets of genetic counseling is informed consent. Our job is to provide enough information so families can make an informed decision. We help walk through the benefits and risks of genetic testing options.

Why is genetic counseling essential in healthcare?

A: Genetic counselors can take the time to explain the nuances of genetic testing. For example, sometimes tests can come back with uncertain results or can have incidental findings. Genetic testing is more complicated than general clinical testing, and in our roles, we have the time and skills to navigate that landscape with families. We can also explain the impact of test results on the individual as well as potential impacts on other family members.

Our Staff

MEDICAL

Amy Albright, MS, CGC | *Genetic Counselor*
Vincent J. Carson, MD | *Pediatric Neurologist*
Jennifer Giacoio, CMAA | *Patient Navigator*
Julia A. Goroff, DO | *Pediatrician*
Candace Kendig, RMA | *Practice Manager*
Grace L. Meier, MD | *Family Medicine Physician*
Laura Poskitt, DO | *Medical Director*
Peggy Riehl | *Medical Receptionist*
Dawn Sheets, GCA, CMAA | *Genetic Counseling Assistant*
Susan Walther, MS, CGC | *Genetic Counselor*

NURSING

Keturah Beiler, BSN, RN, CHPPN
Cherished Lives Program Manager
Christine Hendrickson, RN, BSN, PED-BC
Outreach Program Manager
Alexis McVey, BSN, RN, CPN | *Nursing Director*
Andrea Patel, RNC-NIC | *Nurse*
Donna L. Robinson, MSN, CRNP | *Nurse Practitioner*
Anne Thomas, LPN | *Licensed Practical Nurse*
Sarah Thomas, RMA | *Medical Assistant*

LABORATORY

Cara Forry | *Laboratory Scientist I*
Alanna Koehler, PhD | *Assistant Laboratory Director*
KaLynn Loeven | *Laboratory Scientist II*
Erik G. Puffenberger, PhD | *Laboratory Director*
Sean Schreckengast | *Laboratory Scientist I*

RESEARCH

Karlla W. Brigatti, MS, CGC | *Research Director*
Joelle Williamson Clark, MPH
Clinical Research Manager
Ashlin Rodrigues, MS | *Clinical Research Analyst*
Kevin A. Strauss, MD
Head of Therapeutic Development
Erin Sweigert | *Research Associate*

DEVELOPMENT

Skye Gawn | *Development Associate*
Renny Magill, CFRE | *Development Director*
Julia Martin | *Development Associate*
Kelly Woodson | *Event Manager*

ADMINISTRATION

Kimberly Broadbent | *Accounts Payable Clerk*
Kelly Cullen | *Marketing & Communications Manager*
Adam D. Heaps, MS, MBA | *Executive Director*
Jessica Snyder, PHR, SHRM-CP
Human Resources Generalist
William Van Ess, MS, CFE | *Accounting Manager*

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Stephen Tifft, MD
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Glenn Zimmerman

Pictured: A blood spot sample being prepared to run through an HPLC machine

Matching Grant Opportunity

To fund critical lab equipment

The Clinic provides testing of amino acids through our in-house laboratory, which is critical for patients with metabolic diseases we commonly treat, including maple syrup urine disease (MSUD), phenylketonuria (PKU), and glutaric aciduria type 1 (GA1). Our lab currently has two instruments that run these tests, as many as 14 per day, so that we can provide same-day results. This significantly improves patient treatment plans and avoids life-threatening complications.

One of the two is nearing the end of its useful life, and now, thanks in part to an anonymous $20,000 matching grant, we are launching fundraising for the $80,000 needed to replace this instrument, called an HPLC (high-performance liquid chromatography). If you are interested in learning more about helping us fund this important laboratory equipment, please contact our Development team.

Did you know?

You can automate your giving to the Clinic. Generous donors who prefer the ease of monthly giving can select to have a monthly amount debited from their bank account or have their credit card billed monthly. To learn more about this option, please contact our Development team or visit our website and click on 'Give'.

You can support a special project at the Clinic. Thanks to the generosity of our community, more than 70% of the Clinic's revenue comes in the form of general charitable support. This form of support ensures that the Clinic can provide critical clinical and laboratory services and cutting-edge research while keeping fees affordable for patients. In addition, the Clinic receives gifts for special projects that fund specific research, laboratory projects/equipment, and programs like our Cherished Lives palliative care program. For example, we are grateful for donors, who through the Anabaptist Foundation have donated just over $250,000 since the beginning of 2025 to support two Clinic programs; Cherished Lives and the Plain Insight Panel™ test. These donations significantly fund these two critical projects and allow the Clinic to pursue these additional services that support patients and their families. For information, please contact our Development team.

You can make a future gift to the Clinic through your estate plan. Through your will, a charitable trust, or simply designating the Clinic as a beneficiary of your IRA or life insurance plan, you can support the Clinic's work and leave your legacy. Often, these gifts are relatively simple to do. If you have questions about your options, please contact our Development team.

Development Team Contact Information

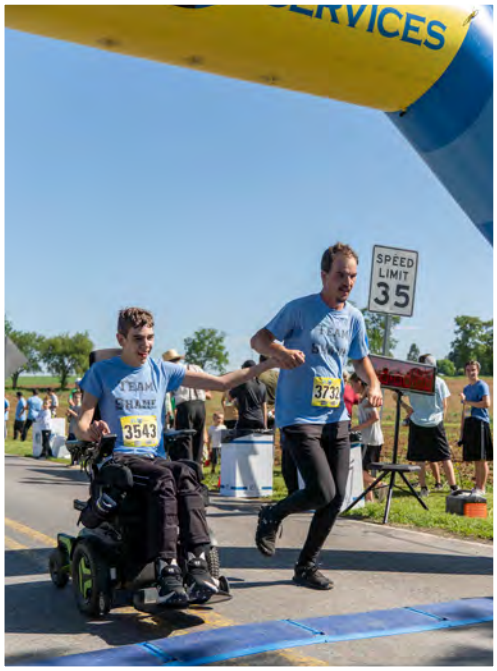
Renny Magill, Development Director
rmagill@ClinicforSpecialChildren.org or 717-687-9407

Clinic for Special Children 5k

May 17, 2025 | Leola, PA

On Saturday, May 17, 2025, we hosted a record-breaking 5k at the Leola Produce Auction! Over 445 participants and 30 sponsors raised over $70,000 to support the Clinic. Thank you to everyone who participated, the volunteers who made the event happen, and our generous sponsors!

A special thank you to Nemours Children's Health, our Presenting Sponsor since the race began in 2018! As featured in past newsletters, Nemours is a long-standing collaborator and supporter of the Clinic. We look forward to next year's race!



Clinic *for* Special Children®

PO Box 500
Intercourse, PA 17534

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

ClinicforSpecialChildren.org

Genetic Counseling at the Clinic

Read a patient's story about their journey to a diagnosis, meet our genetic counselors, and learn more about genetic counseling in this edition of our Clinic newsletter.

2025 Benefit Auctions

We're already halfway through our 2025 benefit auction season! Join us at an upcoming auction in Ohio, Missouri, Pennsylvania, or New York - learn more inside!

The Clinic's Mission

Improving the quality of life for those with genetic or complex medical conditions through innovative research, accessible laboratory services, and compassionate care.