



NICU & MSUD

The Zimmerman Family

When Melissa Zimmerman heard a family member exclaim that her newborn baby girl smelled 'so sweet', she immediately knew something was amiss. Her newborn daughter, Kourtney, became fussier with each passing day and was refusing to eat. "After seeing how she was acting and hearing someone say that she smelled sweet, I had a gut feeling that she had Maple Syrup Urine Disease (MSUD)," explains Melissa. "We called the Clinic immediately and just two hours later, we were headed to Lancaster General Hospital."

Even though it was a weekend, the Zimmerman family was greeted at Penn Medicine Lancaster General Hospital by Dr. Vincent Carson (Pediatric Neurologist) and Donna Robinson (Nurse Practitioner) from the Clinic.

"The Clinic team met us in the parking lot of the hospital to make sure we didn't go through the emergency room with Kourtney. There they took a blood sample for urgent genetic testing to confirm she had MSUD. Donna even sampled Kourtney's earwax to see if it exhibited the classic sweet smell of a baby affected with MSUD, and it did." MSUD is a serious inherited metabolic disorder in which the body cannot process certain amino acids. If untreated, brain swelling can occur which can cause irreversible brain damage or death.

With her serious condition and the severity of her uncontrolled MSUD, the Clinic's doctors admitted Kourtney to Penn Medicine Lancaster General Health's Women and Babies Hospital (WBH). For the next 17 days, Kourtney was cared for around the

clock at WBH's NICU (neonatal intensive care unit). "The NICU team at WBH was so great and helpful. They worked hand-in-hand with the Clinic's providers to educate us on her rare condition, and reassure us that everything would be okay," explains Melissa.

"Women and Babies Hospital has Lancaster County's first and most advanced NICU in our region. When we have to admit newborns facing serious and rare conditions, their team works collaboratively with us to ensure the child receives the specialized care they need. Other times, they will call the Clinic to help with a baby in need of specialized genetic testing. This partnership has elevated the care of many newborns. We are lucky to have them in our backyard here in Lancaster County," says Dr. Laura Poskitt, Medical Director at the Clinic.

"It was quite a journey during those early days with Kourtney. We had to watch her regain skills, like drinking from a bottle, that she lost while she was sick. We are grateful for the level of care WBH's NICU provided our family and how they worked with us. They are above and beyond," says the Zimmerman family. "With the care she received, I don't think she has any lasting damage and she continues to surprise us every day."

Today, Kourtney is just like any other 18-month-old. She's strong and tries to do everything that her siblings can do! She currently visits the Clinic every two months for checkups and will soon be put on the list to receive a liver transplant for her MSUD.



SPECIAL DATES

Community Benefit Dinner Tuesday, October 1

Martindale Fellowship Center | Ephrata, PA

Finger Lakes Benefit Auction Saturday, October 5

Ontario Produce Auction | Stanley, NY

New Holland Food Truck Festival October 4 & 5

Country Lane Gazebos | New Holland, PA

The Extraordinary Give Friday, November 22

All Day | Online Fundraiser

www.ExtraGive.org

Thanksgiving - Office Closed November 28 & 29

Closed all day

Christmas Eve - Office Closed Tuesday, December 24

Office closed from 1 p.m. - 5 p.m.

Christmas - Office Closed December 25 & 26

Closed all day

New Year's Eve - Office Closed Tuesday, December 31

Office closed from 1 p.m. - 5 p.m.

New Year's Day - Office Closed Wednesday, January 1, 2025

Closed all day

Good Friday - Office Closed Friday, April 18, 2025

Closed all day

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



Staff News

Julia A. Goroff, DO



We're delighted to welcome Dr. Julia A. Goroff to the Clinic!

During her residency, she spent time at the Clinic and shared our values in the advancement of health care for children and research.

She joins our team as a Pediatrician and works as a member of our clinical team to evaluate, diagnose, and treat children who visit the Clinic for care.

Dr. Goroff and her family recently moved to Lancaster from the Philadelphia area and are looking forward to learning more about their new home!

Laura Poskitt, DO



Congratulations to Dr. Laura Poskitt, Medical Director at the Clinic, for celebrating her 5th work anniversary!

Dr. Poskitt joined the Clinic in July 2019 as a Pediatrician and was promoted to our Leadership Team as Medical Director earlier this year.

As our Medical Director, Dr. Poskitt manages our clinical operations, oversees our physician team, and develops and implements strategic goals to fulfill the clinical mission of our organization.

Board Updates

Jan L. Bergen



Jan L. Bergen is retiring from the Clinic's Board of Directors in October. Jan joined the Clinic's board in 2021 and has been instrumental in many of

the collaborations between the Clinic and Penn Medicine Lancaster General Health. She was a

vital member of our new building fundraising committee and an advocate for the Clinic in the greater Lancaster community. We're grateful for Jan's guidance and leadership!

Leonard Hurst

After serving for nearly 30 years on the Clinic's Board of Directors, Leonard Hurst will be retiring in October. Leonard has been a part of the Leola Auction Committee since its inception and has been a guiding light for the Clinic over many decades.

We're deeply grateful to Leonard and his family for their dedication to the Clinic over many years.

Glenn Zimmerman

We are excited to welcome Glenn Zimmerman to the Clinic's Board of Directors! Glenn was a member of the Clinic's capital campaign committee and helped raise funds for our new building project. He is employed as the Lititz store manager as part of his family's business, PaulB Hardware.

He lives in Stevens, PA, and has four children with his wife, Anita. He has church and family members who have benefitted from the Clinic's services and is passionate about the Clinic's work. "Seeing the families that have children going to the Clinic and the improved lives they can live with the Clinic's help is inspirational. We give all praise and honor to God who has helped the Clinic with this good work," shares Glenn.

2025 CSC 5k

The Clinic for Special Children's 5k is coming back in the spring! We plan to release more details in early 2025. Stay tuned for information on how to register, the course, and much more.

If you're interested in being a corporate sponsor or volunteering at our 2025 race, please contact our development team at giving@clinicforspecialchildren.org or 717-687-9407.

It's Flu Shot Season!

It's that time of year when the bugs start go-

ing around! Please call the Clinic at 717-687-9407 to schedule your or your child's flu shot. Anyone over six months of age is encouraged to get a flu shot each year. We offer flu shots to our patients and their immediate family members.

The best way to protect yourself and your family during the flu season is to avoid exposure, practice good handwashing, and receive your annual flu shot.

Development Committee Volunteers Needed!

We are looking for two volunteers to serve on the Clinic's development committee. The development committee meets quarterly for a couple of hours and provides guidance on fundraising and marketing to the Clinic's staff and board of directors.

For one volunteer position, we are looking for an experienced development professional who has served in the greater Lancaster community. Ideally, this person would have experience with major gifts, planned giving, event management, grant writing, and overall fundraising best practices.

For the second volunteer position, we are looking for an experienced marketing or communications professional. This person would have experience in advertising, public relations, communications, graphic design, and more.

If interested, please contact Adam Heaps, MS, MBA, Executive Director at 717-687-9407 or aheaps@clinicforspecialchildren.org.

New Holland Food Truck Festival Hosted by Country Lane Gazebos

We're excited to be a part of the New Holland Food Truck Festival on Friday, October 4th (11 am - 7 pm) and Saturday, October 5th (9 am - 3 pm) at Country Lane Gazebos (540 Hollander Road, New Holland, PA)!

Several activities will benefit the Clinic, and we will have an informational table to share about our work! Bingo benefitting the Clinic will start at 9:30 a.m. on Saturday. Stop by to enjoy balloon art, pony rides, lumber maze, face painting, delicious food, and more!

RECENTLY PUBLISHED PAPERS

Melissa O. Bowman, MD, Devyani Chowdhury, MD, **Laura E. Poskitt, DO**, Aaron Chidekel, MD. **High-tech care in the Amish: Chronic respiratory failure in Ellis-van Creveld syndrome.** Pediatric Pulmonology. 2024 Jun 18. doi: 10.1002/ppul.27139

Susan E. Matesanz, **Karlla W. Brigatti**, Millie Young, Sabrina W. Yum, **Kevin A. Strauss**. **Preemptive dual therapy for children at risk for infantile-onset spinal muscular atrophy.** Annals of Clinical and Translational Neurology. 2024 May 31. doi: 10.1002/acn3.52093

Michelle Dookwah, Shannon K Wagner, Mayumi Ishihara, Seok-Ho Yu, Heidi Ulrichs, Michael J. Kulik, Nadja Zeltner, Stephen Dalton, **Kevin A. Strauss**, Kazuhiro Aoki, Richard Street, Michael Tiemeyer. **Neural-specific alterations in glycosphingolipid biosynthesis and cell signaling associated with two human ganglioside GM3 synthase deficiency variants.** Human Molecular Genetics. 2023 December 1. doi: 10.1093/hmg/ddad146

Sunil Muthusami, MD, MBBS, Laura Hatchman, MD, **Vincent J. Carson, MD**. **Hypotonia and failure to thrive in an 8-month-old infant.** Pediatrics in Review. 2023 Nov 1. doi: 10.1542/pir.2021-005040

*bolded names indicate current CSC staff authors

EXTRATM GIVE



LANCASTER COUNTY
COMMUNITY
FOUNDATION



MURRY
FOUNDATION

Friday, November 22 ExtraGive.org

It's almost that time of year again! The **Extraordinary Give** - Lancaster County's largest day of online giving - is on **Friday, November 22nd**! Help the Clinic **raise \$75,000 in 24 hours** for our mission. You don't need to be located in Lancaster County - we have donors from all over the United States who support the Clinic through the ExtraGive.

Every dollar you donate on November 22nd during the ExtraGive will be stretched by a pool of more than \$500,000 from local sponsors.

Donating is simple - here are **three ways you can support**:

- 1 On Friday, November 22nd (from midnight to 11:59 p.m.) **visit www.ExtraGive.org** and search 'Clinic for Special Children' to give online on our page during the 24-hour period.
- 2 **Call the Clinic at 717-687-9407** between 9:00 a.m. - 5:00 p.m. on Friday, November 22nd to donate via credit card over the phone.

Won't have access to the internet on the day of ExtraGive? Call us before and we'll help process your ExtraGive donation for you on the day of ExtraGive.
- 3 **Set up your fundraising page!**
You can create your own fundraising page to share with your friends and family to raise money for the Clinic! Visit www.ExtraGive.org, search for 'Clinic for Special Children', and click the 'Fundraise' button to create your page before November 22nd!

Scan the QR code
for a shortcut to
our giving page!



Meet our Providers

We're fortunate to have a great team of knowledgeable, compassionate, and caring providers. We asked them to share more about their work and the Clinic!



Julia A. Goroff, DO
Pediatrician

Why are you passionate about the Clinic's work?

I had the fortune to spend time at the Clinic during my Pediatric residency. During that time, I witnessed the dedication to providing excellent care and the pursuit of research

to advance that care. Yet, what struck me the most and made me so excited to join the Clinic was the trust and mutual commitment the families and providers have to manage their child's care and the health of their community and future generations. I am honored to join the Clinic and community in this partnership!



Grace L. Meier, MD
Family Medicine Physician

What do you wish people knew about the Clinic & our work?

I wish people knew how selfless and forward-thinking patients of the Clinic can be, especially in regards to trying new therapies and enrolling in clinical research trials.

They're paving the way to access novel treatments for a multitude of rare diseases that affect people all over the world.

What's your favorite part of your job?

My favorite part is being creative in how to help patients. I love a good challenge. It's rewarding to help them find solutions.



Donna Robinson, MSN, CRNP
Nurse Practitioner

What makes the Clinic a special place?

We try to maintain a personal touch with all our patients and their families. The Clinic works with the patient & family to be a team. We advocate for patients with billing costs and finding patient assistance programs.

What's your favorite part of your job?

I enjoy the people I work with, both staff and patients. From a work standpoint, I enjoy metabolic diets and looking at the amino acids, performing procedures such as IV infusions, patient teaching, g-tube changes, etc.



Vincent J. Carson, MD
Pediatric Neurologist

Why are you passionate about the Clinic & why did you choose to work here?

To me, the Clinic represents how medicine should be practiced. We are focused on finding pragmatic answers for people who have difficult problems.

The Clinic is focused on being precise, both in diagnosis and treatment. The Clinic demonstrates how you can provide a high level of quality care without the exorbitant cost of modern healthcare.



Helge Hartung, MD
Physician-Scientist

Why are you passionate about the Clinic & why did you choose to work here?

I am inspired by the fact that our Clinic is here to serve the community, as opposed to the community serving the Clinic. My goal is to leverage my long-standing experience in academic medicine

so that our Clinic can prosper and continue to grow its precious mission. As the first Physician-Scientist at the Clinic, I look forward to helping map out new clinical and research needs and opportunities and be a part of the Clinic for this exciting phase of the organization's history.



Laura Poskitt, DO
Medical Director

What do you wish people knew about the Clinic & our work?

Many people know about the medical care we provide for the community but are not aware of how our laboratory and research departments are intertwined with the care we give. Our

decades of cutting-edge research into rare diseases have allowed us to be a part of many medical breakthroughs locally and globally, making a difference in so many lives. Our innovative lab can obtain a genetic diagnosis on at-risk newborns faster than almost anywhere in the world, preventing death and disability in many children.



Collaborator Spotlight

Katie A. Preedy, APRN-BC
Nemours Children's Hospital, Delaware

The Nemours Children's Hospital in Wilmington, Delaware, offers a Level IV NICU, the highest level of neonatal care. The Clinic for Special Children has frequently collaborated with the Nemours NICU team to ensure infants facing serious rare genetic disorders get the best care possible. Katie Preedy, APRN-BC, the manager of the Neonatal Advanced Practice Provider (APP) team at Nemours, is a close collaborator of the Clinic and has provided compassionate care to many Clinic families.

Raised in Dresher, PA, Katie has spent most of her career working as a Neonatal Nurse Practitioner (NNP) at Nemours. She attended West Chester University, studied nursing, and earned her degree. She later moved to California and worked as a travel nurse, where she would later meet her husband, Ian. For a year, they took a hiatus from working to backpack and camp throughout New Zealand, Australia, and Southeast Asia. After returning home, she went to school to earn her master's degree as an NNP at the University of Pennsylvania.

"I learned about the Clinic more than a decade ago while caring for a baby with CODAS syndrome in our NICU. I was unfamiliar with this rare condition and learned about the capabilities and scope of the state-of-the-art on-site laboratory at the Clinic, which allows the clinicians to quickly diagnose genetic disorders that might otherwise take weeks or months at other centers," explains Katie. "Over the years, the expertise and communication from the Clinic staff has helped our NICU team understand more about some of the genetic conditions affecting the unique population we care for. We've also cared for Clinic families in our Advanced Delivery Unit (ADU), which allows expectant mothers carrying infants with prenatally diagnosed congenital anomalies to deliver at Nemours, where the babies can be closely monitored before they are born and immediately after birth to optimize their outcomes."

In addition, Katie has collaborated with the Clinic team and other Nemours physicians on research and co-authored a publication to evaluate clinical outcomes of patients with Ellis-van Creveld syndrome (EVC). "I have a clinical interest in caring for babies with skeletal dysplasia and other rare conditions," she shares. "Through this research and the col-

laboration of the Clinic and the NICU team at Nemours, it's helped us implement some condition-specific practices in the NICU, which we feel has improved the care they receive and shortened the hospital length of stay for many patients from the Plain Community." Katie has presented at several educational and family days hosted by the Clinic and serves as a liaison between the Clinic and Nemours to make the discharge process to home a smooth transition.

"Serving as a liaison between the Clinic and Nemours has allowed me to get to know many Clinic families. It has been a privilege to visit some families in their homes, farms, and even at their children's schools. Seeing families in their environment helped me better understand their lifestyle and practices so we can support them with everything they need to be safely discharged home," shares Katie.

"Katie has been a long-time advocate for Plain families at Nemours. It is common for Katie to call us with a baby just born to a Plain family," shares Dr. Laura Poskitt, the Clinic's Medical Director. "We often work together to find a genetic diagnosis quickly, inform the family, work with the NICU team to customize the care plan and collaborate to ensure the baby comes home as soon as possible. Since Katie has ensured the Clinic has been involved all along, we can provide the most seamless care transition from hospital to home."

When asked about the proudest accomplishment of her career, Katie says, "the privilege of working with a team of exceptional medical professionals and an entire discharge team who have a common goal of delivering the safest and best care possible to infants and children while also supporting each other. From making sure these vulnerable infants have the best possible start in life to sending them home with parents who are prepared to care for them safely has been my professional mission."

Katie and her husband share three sons: Evan, Luke, and Jack. She enjoys camping, hiking, pickleball, and anything outdoors. We're grateful to Katie and the Nemours team for their dedication to providing the highest level of care to babies in need.

Our Staff

Amy Albright, MS, CGC | Genetic Counselor

Keturah Beiler, BSN, RN, CHPPN
Nurse & Cherished Lives Program Manager

Karla W. Brigatti, MS, CGC | Research Director

Vincent J. Carson, MD | Pediatric Neurologist

Joelle Williamson Clark, MPH
Clinical Research Manager

Kelly Cullen | Marketing & Communications Manager

Skye Gawn | Gift Processing & Data Entry Coordinator

Jennifer Giacoio, CMAA | Medical Receptionist

Julia A. Goroff, DO | Pediatrician

Helge Hartung, MD | Physician-Scientist

Adam D. Heaps, MS, MBA | Executive Director

Christine Hendrickson, RN, BSN, PED-BC | Nurse

Candace Kendig, RMA | Office Manager

Alanna Koehler, PhD | Assistant Laboratory Director

KaLynn Loeven | Laboratory Scientist II

Julia Martin | Development Associate

Alexis McVey, BSN, RN, CPN | Nursing Director

Grace L. Meier, MD | Family Medicine Physician

Laura Poskitt, DO | Medical Director

Erik G. Puffenberger, PhD | Laboratory Director

Donna L. Robinson, MSN, CRNP | Nurse Practitioner

Ashlin Rodrigues, MS | Clinical Research Analyst

Sean Schreckengast | Laboratory Scientist I

Dawn Sheets, CMAA | Medical Receptionist

Jessica Snyder, PHR, SHRM-CP
Human Resources Generalist

Kevin A. Strauss, MD
Head of Therapeutic Development

Erin Sweigert | Research Associate

Anne Thomas, LPN | Licensed Practical Nurse

Sarah Thomas, RMA | Medical Assistant

William Van Ess, MS, CFE | Accounting Manager

Susan Walther, MS, CGC | Genetic Counselor

Kelly Woodson | Event Manager

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Herman Bontrager | Chair of the Board

Elam Esh

Leon Hoover | Vice Chair

Mark Martin | Treasurer and Chair-Finance Committee

Jacob Petersheim

Jonathan H. Salvin, MD

Stephen Tifft, MD

Glen Zimmerman | Chair-Development Committee

Glenn Zimmerman



Donor Spotlight

Swing Kingdom

One of the first things people (especially children!) notice as they approach our new Clinic for Special Children building, is a fully accessible playground welcoming them by our entrance. The Clinic's new playground is one of the most exciting features of our new facility, and it's all thanks to the team at Swing Kingdom in Leola, PA. They generously donated the fully ADA and wheelchair-accessible playset to the Clinic's new building project, free of charge.

Started on an Amish farm in Lancaster County in 2000, Swing Kingdom is the largest manufacturer of vinyl playsets in the United States and is headquartered in Lancaster County. From infant swings to playhouses, they have designed solutions for playset needs in residential and commercial settings. Throughout three decades of growth and changes, their family-operated business has always strived to keep God, quality, value, and customer service at the forefront of what they do. Their motto, 'mighty playsets, serious fun', hints at their desire to see every family enjoying serious fun in their backyard, church, school, park, or clinic!

When the team at Swing Kingdom heard of the Clinic's need for a playground at our new facility in Gordonville, they jumped at the opportunity to help. "Knowing the work that the Clinic does for children in need not only in our local community, but around the globe, and the impact their work has on young lives, we felt we wanted to be a part of the project," says Amy Martin, Sales Director at Swing Kingdom. "The family at Swing Kingdom is grateful for the opportunity to be part of the work being done at the Clinic. Our owner learned about the Clinic through the local community and some of our employees have family members who use or have used the Clinic's services."

By being in the business to 'bring fun and joy to families', the choice to provide a fully accessible playground for the Clinic was a natural fit for Swing Kingdom. "What better way to support the work being done at the Clinic than to help bring joy to children visiting for appointments and giving them something to look forward to - enjoying a playset built especially for them," shares Amy. "We hope that through the playset being right there at the entrance for children, they will be able to enjoy their visits more and maybe even look forward to coming in for a visit. Our prayer is that we too can make a difference in their lives both at the Clinic and at home."

Since the Clinic opened its doors in Gordonville earlier this year, countless children of all abilities have enjoyed the new playset space. At the Clinic, we are deeply grateful to the entire Swing Kingdom team for helping us bring joy to families who visit us for care and for making sure every child can play at the Clinic.

THANK YOU

to everyone who joined us at a benefit auction this year! Enjoy a roundup of photos from this year's auction season. A special thank you to the many volunteers who organize the auctions every year - we greatly appreciate your support!

2024 Benefit Auction Season

Food, fun, & fellowship!





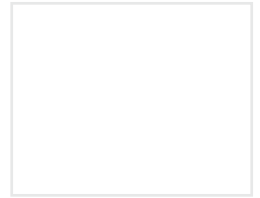
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2024 Extraordinary Give

Help us raise \$75,000 in just 24 hours!

Save the date for Lancaster County's largest day of online giving on Friday, Nov. 22nd! Details inside on how to help the Clinic fundraise during the ExtraGive.

Rare Genetics in the NICU

Read about how we partner with local hospital NICU teams on treating infants facing rare genetic disorders and learn about a family's journey with Maple Syrup Urine Disease (MSUD) in the NICU.

The Clinic's Mission

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