

Newsletter

ISSUE #53 SUMMER 2021



Adult Services at the Clinic for Special Children Katie Martin

Katie Martin has experienced seizures for as long as she can remember. "When I was two years old, my family thought I was having nightmares when they would hear me in the middle of the night. It was only when I started having seizures during the day that we realized they were actually seizures." Throughout her childhood, Katie tried many different medications to reduce her seizure activity – all to no avail.

Over the years Katie had heard about the Clinic for Special Children through family and friends. Even though she was placed on the medication Lamotrigine by her neurologist in her early 20's with some success, she decided last year to give the Clinic a try.

Katie's timing was perfect. The Clinic had recently hired Dr. Grace Meier (formerly Dr. Loudon), a Family Medicine Physician, to expand services to care for adult patients. At Katie's first visit to the Clinic, she met with Dr. Meier to discuss her history of seizures and a nagging abdominal pain. Dr. Meier quickly ordered an epilepsy panel to test Katie for a variety of genetic conditions that can cause seizures.

Katie's test came back positive for NPRL3-related epilepsy. It just so happens that the NPRL3 genetic variant was discovered by the Clinic in 2016 to be a cause of epilepsy. Katie's diagnosis of NPRL3-related epilepsy provided

her with the cause of her seizures and may help guide the management of seizures in the future.

To address Katie's abdominal pain, Dr. Meier referred her to a specialist to get an ultrasound scan. The ultrasound results showed that Katie needed surgery, which Dr. Meier worked to coordinate with a surgeon. The surgeon removed a nine centimeter endometrioma and Katie felt relief almost immediately.

In addition to *NPRL3*-related epilepsy and the endometrioma, Dr. Meier noticed a history of elevated blood sugars and drew attention to the importance of managing her pre-existing diabetes. Katie now works with a nutritionist and gets regular blood test checkups.

When asked about what adult services at the Clinic means to her, Katie exclaims, "I'm happy to hear that the Clinic is offering adult services. Your team understands and works with conservative cultures and helps keep prices low for your patients. I'm grateful to Dr. Meier for working with me on the management of my seizures and pre-existing diabetes. I'm thankful that I found the Clinic for Special Children and Dr. Meier."

Today Katie enjoys working at Uncle Henry's Pretzel Bakery and visits the Clinic a couple of times a year for management of her seizures and surveillance of her diabetes.

SPECIAL DATES

Missouri Benefit Auction Saturday, August 21 Ed Good Family Farm

Ed Good Family Farm Memphis, MO

Labor Day - Office Closed Monday, September 6 All Day

Blair County Benefit Auction Saturday, September 11

Morrison's Cove Produce Roaring Spring, PA

Clinic for Special Children 5k Saturday, September 18

At the Clinic

9 a.m. - 11 a.m.

Application and details inside!

Community Benefit Dinner Wednesday, October 13

Martindale Fellowship Center 4 p.m. - 7:30 p.m.

Details inside!

2021 Extraordinary Give Friday, November 19

Online Fundraiser - All Day
Help the Clinic fundraise in 24 hours by
spreading the word with your family and
friends!

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

Staff News Amy Albright, MS



We're excited to welcome Amy Albright to the Clinic!

Amy will work as a Genetic Counselor at the Clinic and assist in new patient triage and

intake, provide genetic counseling services, and manage logistics of the molecular diagnostics service.

Amy recently graduated from Thomas Jefferson University with her Master's of Science in Human Genetics and Genetic Counseling. She grew up in Lancaster County and graduated from Hempfield High School.

In her free time, she enjoys doing anything outside (like hiking!) and spending time with her beloved cat.

Emilienne (Emi) Bolettieri



We're excited to welcome Emilienne (Emi) Bolettieri to the Clinic!

Emi will work as a Research Associate and will support a number

of the Clinic's research endeavors, including work on the Phenylketonuria (PKU) and TNNT1 myopathy projects.

Emi recently graduated from Cornell University with a degree in Biology and minors in Viticulture & Enology, Creative Writing, and Nutrition & Health.

In her spare time, she enjoys weight training, creative writing, and spending time with friends and family.

Candace Kendig, RMA



Congratulations to Candace Kendig, RMA who recently earned her certification as a Registered Medical Assistant (RMA)!

Candace joined the

Clinic in 2017 and works as our Office Manager. She is currently pursuing a Bachelor's Degree in Healthcare Management from Central Penn College with an anticipated graduation in Fall of 2021.

Candace enjoys positively impacting the children and families living with rare genetic disorders that visit the Clinic.

Grace Meier, MD



Congratulations to Dr. Grace Meier, formerly Dr. Loudon, who recently got married in May!

Moving forward, she will be going by her

new married name, Dr. Meier. While her name has changed, Dr. Meier is still the friendly and familiar face many of you know as our Family Medicine Physician at the Clinic.

Emily Seitz, PhD



Congratulations to our Development Director, Dr. Emily Seitz! Dr. Seitz recently successfully defended her dissertation and earned her doctorate from the

Pennsylvania State University.

Dr. Seitz deeply enjoys connecting funders and ideas and finding support for community health initiatives at the Clinic.

Kevin A. Strauss, MD



In July we celebrated a milestone work anniversary for our Medical Director, Kevin A. Strauss, MD! Dr. Strauss celebrated his 20th anniversary at

the Clinic for Special Children!

Dr. Strauss has co-authored over 60 peer-reviewed journal articles and has given many lectures around the globe about the important work at the Clinic. He joined the Clinic team in 2001 as a Pediatrician and was promoted to Medical Director in 2008.

Thank you for your dedication to the Clinic, patients, and their families!

Important COVID-19 Updates

Vaccine

We have reviewed the available data from studies on the authorized COVID-19 vaccines, which show the vaccines are both effective and safe.

The COVID-19 vaccine is now available for individuals who are 12 years old or older. Please contact your doctor with questions regarding the COVID-19 vaccine. If you are interested in being vaccinated, please call the national vaccine hotline at 1-800-232-0233 or visit Vaccines.gov for more information.

For individuals near Lancaster County, you can contact the WellSpan Health COVID-19 hotline at 855-851-3641. For information on Lancaster County's available vaccine locations, visit VaccinateLancaster.org or call 717-588-1020.

Office Operations

We continue to require mask wearing for all visitors to our clinic facility. If you have a scheduled appointment and are feeling unwell, we ask that you call us at 717-687-9407 before coming to your appointment. Our clinic staff is now back to operating at full capacity, as our full-time staff are fully vaccinated for COVID-19. If you have any questions before your visit, please contact us at 717-687-9407.

Save the date! Community Benefit Dinner

Wednesday, October 13th 4:00 p.m. - 7:30 p.m.

at the Martindale Fellowship Center

We are looking forward to our 2021 Community Benefit Dinner on Wednesday, October 13th from 4:00 p.m. - 7:30 p.m. at the Martindale Fellowship Center (352 Martindale Road, Ephrata, PA 17522)!

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 dinner is by donation only. All proceeds will benefit the Clinic for Special Children's mission.

RSVP's are required to attend. To RSVP, please call the Clinic at 717-687-9407 or email RSVP@clinicforspecialchildren.org.

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4th Annual



5K

Saturday, September 18, 2021 9:00 a.m. - 11:00 a.m.

at the Clinic for Special Children

\$30 (by Aug. 20th, includes goody bag & t-shirt)

\$35 (after Aug. 20th, goody bag & t-shirt while supplies last)

\$10 Kid's color fun run (ages 10 & under)

KID'S COLOR FUN RUN

BAKE SALE

BREAKFAST SNACKS FOR SALE

Register for the Clinic for Special Children 5k! You can register by:

- 1) Filling out the application included in this newsletter *OR*
- 2) Visiting www.ClinicforSpecialChildren.org/events/5k

 OR
- 3) Calling the Clinic at 717-687-9407

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4) Scanning the QR code



Mark your calendars for the remaining

2021 Benefit Auctions

7:00 a.m. | Breakfast 8:30 a.m. | Auctions Begin Physician Remarks and Quilts to Follow

Missouri Auction | August 21

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

Blair County Auction | September 11

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



Please visit our auctions website (www.ClinicAuctions.org) to view the most current auction information.

We recognize the rapidly evolving situation with COVID-19 and have been taking all precautions to keep our staff and patient families safe. Auctions are subject to change due to COVID-19 restrictions.



Adult Services at the Clinic

Interview with Vincent Carson, MD, Clinical Operations Director

Learn more about the Clinic's expansion of adult services below in an interview with our Clinical Operations Director, Dr. Vincent Carson.

What are adult services?

We use the term 'adult services' to refer to the specialized and coordinated care we provide to our adult patients. This care can range from primary care to specialized care. Adult services at the Clinic are the continuation of care that we provide our pediatric patients as they enter adulthood and for the rest of their lives.

At the Clinic, we focus specifically on patients with rare genetic disorders or complex medical conditions that require complex medical care. Our adult services program will continue to focus on adults with special needs who have genetic disorders or complex medical conditions.

Why is the Clinic offering adult services?

When the Clinic was founded, it was initially started to serve children with a handful of genetic or metabolic conditions. Now that the Clinic has been around for over 30 years, we see over 400 conditions and many of the children are now adults who

Current Active Patie<u>nts</u>

33% 14 years or older

25% 21 years or older still require specialized care. Many physicians are uncomfortable caring for some of the rare genetic conditions that we see, and we realized that we needed to develop the expertise of caring for adults in-house at the Clinic.

Many of our patients are from the Plain community and they are self-pay. At the Clinic, we can provide them with the affordable and accessible care that they need.

Many of our patients also have established a lifelong trust with us and prefer to keep their care with the Clinic since we know them and their disorder well.

What changes have been made at the Clinic?

Last year we hired Dr. Grace Meier as our first Family Medicine Physician. Dr. Meier will be working to formalize our adult services program and work with our clinicians to provide care to adult patients.

Before Dr. Meier's hire, all of our providers were trained in pediatrics. In order for us to provide the best care possible to our patients that are now adults, we needed to hire someone with the skill set necessary to care for them throughout their entire life. With the addition of Dr. Meier, we are able to provide care to our patients throughout their entire life.

Why is the Clinic starting adult services now?

Currently 33% of the Clinic's active patients are over the age

of 14 years and 25% are over the age of 21 years. While we see several conditions at the Clinic that are fatal earlier in childhood, the large majority of our patients have conditions in which they will live well into adulthood.

When we see patients at the Clinic, not many "graduate" from the Clinic. Once we start seeing a patient, we



typically continue to care for them indefinitely. When we see new patients at the Clinic, both children and adults, it adds to the growing number of patients that we see and our volume of patients is continuously increasing.

Over the past two years, we've hired both Dr. Laura Poskitt, a trained Pediatrician, and Dr. Grace Meier, a Family Medicine Physician, to help us meet the ever growing need of the community. We are grateful for the support of the community which allows us to spend time with patients and give them the best care possible. The need for adult services at the Clinic has been a long-time coming, and the need is now.

What does the future look like for adult services?

With the additions to our team, we can provide specialized care across the lifespan.

While the majority of our patients have genetic disorders that start in childhood, we are expecting to see adults that come to our practice with adult onset genetic conditions. In the past, we may not have seen these conditions since we have focused primarily on pediatrics.

We're looking forward to expanding the specialty services that are offered at the Clinic to fit the needs of our adult patients. We'll continuously look to grow our services in a manner that allows us to provide the best comprehensive care to the patients we care for, no matter their age.

Who can come to the Clinic as an adult patient?

We will continue to select patients to come to the Clinic for our services based on the information they provide to us via a new patient packet. Our team will review the information and determine if we are the best fit.

We only accept patients that we feel we can make a difference in their lives with our expertise. If they could be better served elsewhere, we'll make sure to connect them with the right people.

If you're interested in more information on adult services, please call us at 717-687-9407 and we can assist you.



Collaborator Spotlight

Stacy L. Chubb, CRNP, Nurse Practitioner at Compassion Parochial Clinic, CSC Mifflinburg Outreach Clinic

The Clinic for Special Children (CSC) started an outreach clinic in Mifflinburg, PA in 2019 at the Compassion Parochial Clinic (CPC) to see patients closer to where they live. Stacy Chubb, CRNP, a Nurse Practitioner at CPC works closely with our clinicians when they visit the outreach clinic to provide the best care possible to patients in the Mifflinburg area. The CPC is a primary care, self–pay clinic located in the heart of central PA that primarily focuses on the Amish and Mennonite communities.

Stacy always knew that she wanted to be a nurse. In high school, she attended a vocational technical school, focusing on nursing science and earning a certificate as a Certified Nursing Assistant (CNA). After graduating high school, she started taking classes at Harrisburg Area Community College earning an associate degree in nursing and becoming a Registered Nurse (RN). She then worked in nursing at a local hospital, medical-surgical unit, and in corrections (jails and prisons). For 13 years, she worked on a postpartum/maternity unit and earned a Bachelor's of Science degree in Nursing (BSN) from Eastern University and a Master's of Science degree in Nursing (MSN) from Widener University.

During her graduate studies, she had the honor to learn alongside Dr. Mandy Maneval, who introduced her to practicing medicine in the Plain community. Through working as a Family Nurse Practitioner in the Plain community, she was introduced to the CSC. After working for several years in family practice, she heard about the CPC in Mifflinburg, PA. Stacy started working part-time at CPC in 2019 and in 2020 accepted a full-time position as a Family Nurse Practitioner. Stacy explains how her career has evolved, "While growing up my goal was to be a midwife, but after years of hospital nursing and continuing my education, I realized family practice in a rural community for an underserved population is what I found to be the most rewarding. I am humbled to be providing care to the Mennonite and Amish communities."

Stacy is the only full-time provider at CPC and she cares for patients of all ages. She has an amazing collaborator, Dr. Elam Stoltzfus, who is available for any questions she may have and visits the clinic weekly. She cares for patients of every age. Examples of the care Stacy provides includes; getting a call from a local midwife who wants a newborn baby examined, completing a well woman exam including pap testing, helping to set up

end-of-life care for an elderly adult, or collaborating with Dr. Carson to care for a patient with a genetic or metabolic disorder. She also provides urgent care for patients including suturing, injuries, or sick visits. She also does minor procedures in the office including lesion removals, skin biopsies, circumcisions, or casting. "I am always eager to learn new things and hear new ideas to improve the way I practice. I work with amazing nurses and ancillary staff who are very supportive and help keep CPC running day-to-day," explains Stacy.

The CSC team visits Mifflinburg about once a month for our outreach clinic so families do not have to make a long trip to our Strasburg clinic. Stacy supports our team when they visit CPC and acts as the "eyes and ears" closer to home for our Mifflinburg area patients which greatly supports our clinical team at CSC. Stacy also helps collect blood samples for the Plain Insight Panel $^{\rm IM}$ testing at outreach education gatherings within the Mennonite community close to her home.

Stacy greatly enjoys working with the Plain community. "Through the short and unpredictable lives of special children, I have gained many treasured friendships. I have some very close friends within the Amish and Mennonite community. We keep in touch through letters, phone calls, or catching up at auctions. Our families enjoy spending time together for picnics or outings. I have always felt a strong connection to this community by sharing some of the same values and traditions."

Outside of working at CPC, Stacy keeps busy on her horse farm in Oriental, PA. She married her high school sweetheart and has a son and daughter. Her family breeds, raises, and sells quarter horses. Stacy enjoys horseback riding and 'living life through the eyes of her children.' Her son is an avid sportsman; hunting, trapping, fishing, and helping on the farm. Her daughter enjoys horseback riding and outdoor activities. Her family enjoys trips out west to horse auctions or vacationing. She also enjoys reveling in a good book or a nursing/medical journal.

We are so grateful to Stacy for her dedication to patients and their families, especially in the Miffinburg area! She has been a knowledgeable resource for the CSC team as we've established the Mifflinburg outreach clinic and expand into offering adult services.

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Our Staff

Amy Albright, MS Genetic Counselor

Keturah Beiler, RN Nurse & Cherished Lives Program Manager

Emilienne Bolettieri <u>Res</u>earch Associate

Karlla W. Brigatti, MS, CGC Research Operations Director

Vincent Carson, MD Clinical Operations Director

Kelly Cullen Communications Manager

Jennifer Giacoio Medical Receptionist

Adam D. Heaps, MS, MBA Executive Director

Christine Hendrickson, RNC Nurse

Candace Kendig, RMA Office Manager

Lavina King Community Liaison

Caitlin Lavin, MS, MPH, CGC Genetic Counselor

KaLynn Loeven Laboratory Scientist

Julia Martin Development Associate

Grace Meier, MD Family Medicine Physician

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

Emily Seitz, PhD Development Director

Kevin A. Strauss, MD Medical Director

William Van Ess, MS, CFE Accounting Manager

Dawn Wade Medical Receptionist

Millie Young, RNC Research Nurse

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Jacob Zook



Philanthropy at the Clinic How to Support Our Work

You can help ensure cutting-edge compassionate care is accessible to and affordable for everyone. Here's how you can help:

Make a Gift

Consider supporting the Clinic's mission and work through making a gift online at www.ClinicforSpecialChildren.org or through mailing your donation to: Clinic for Special Children, 535 Bunker Hill Road, Strasburg, PA 17579. You can use the included giving envelope in this edition's newsletter to give!

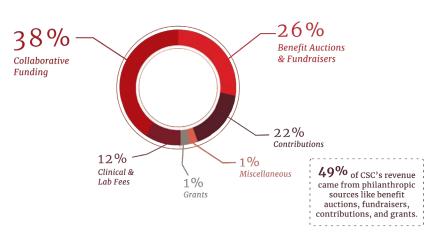
Attend an Event

Attend one of the Clinic's auctions or special events (see upcoming events listed on the right-hand side of the front page of this newsletter).

Participate in Planned Giving

Name the Clinic for Special Children as a beneficiary in your estate plan. For more information, please contact Emily Seitz, Development Director, at eseitz@clinicforspecialchildren.org or at 717-687-9407.

2020 Revenue Snapshot



WHERE ARE THEY NOW?

Four past fellows & students at the Clinic share what they are up to!



Dr. Abigail Benkert

Following graduation from Franklin and Marshall College, I worked at the Clinic for Special Children as the Mary Ellen Avery Fellow from 2013-2014. During this time,

I studied the natural history and treatment options for congenital adrenal hyperplasia. This research project took me across Pennsylvania to patients' homes at all hours of the day. I was welcomed into everyone's home and experienced first-hand the resiliency, advocacy, and graciousness that define the Clinic's patients' families. At the end of my fellowship, I was hired as a full-time laboratory technician and worked in that role until my matriculation at Tufts University School of Medicine in 2015. Throughout medical school, I worked with the Clinic on a number of research projects including investigating the developmental and behavioral aspects of cortical dysplasia focal epilepsy syndrome, as well as studying the natural history of an inherited hypertrophic cardiomyopathy. I graduated medical school in 2019 and am now beginning my third year of training in the cardiothoracic surgery residency program at Duke University Hospital. Even though I work in a large academic medical center now, I carry with me the experiences from the Clinic, which help me focus on delivering patient-centered care. During my research time at Duke, I plan to again collaborate with the Clinic to develop a vector for gene replacement therapy for an inherited hypertrophic cardiomyopathy. My future career goals include pursuing a career in pediatric congenital cardiac surgery, through which I can combine my passion for cardiac surgery with that for genetic disorders and long-term relationships with patients and families.

Lauren E. Bowser

I first joined the Clinic team part-time in February 2017 while I was a student at Franklin & Marshall College. I started as



I left the Clinic in July 2020 to attend the Philadelphia College of Osteopathic Medicine (PCOM) biomedical sciences program. I'm excited to share that I was recently accepted to medical school at PCOM! I'll be starting medical school in August with primary interests in pediatrics, family medicine, and palliative care. Shortly after starting medical school, I'll be getting married to my best friend, Edgar, in September 2021.

My dream is to one day return to the Clinic as a physician to once again serve its mission. The staff at the Clinic and the families they serve have made a tremendous impact on me as a person and as a future clinician and I am so grateful for my time at the Clinic. Some of my favorite memories were made around kitchen tables, as families graciously welcomed me into their homes to participate in one of our research projects. I am better because of the opportunity to have gotten to know their special children.



Mariah Everett

Upon graduating from Swarthmore College in 2018, I interned at the Clinic for a summer working on the NPRL3 epilepsy

project closely with Clinic researchers and clinical staff. I received a Fulbright grant in the Fall of 2018 and spent a year in the Czech Republic teaching English, which was an amazing experience. When I returned to the U.S. I learned that the Clinic had an opening for a Research Associate and I jumped at the opportunity to work with the Clinic team again. I started back at the Clinic in mid-2020 as the Research Associate supporting the Clinic's research efforts and working primarily on the Phenylketonuria (PKU) and WiTNNess studies. Though I spend much of my time working with data, the opportunity to meet with patients and their families and seeing the impact our research can have on their lives has been the most rewarding part of

While I've greatly enjoyed my time at the Clinic, I will be leaving to attend Harvard Medical School this August. I plan to graduate in either 2025 or 2026 depending on if I decide to do a Master's of Public Health. I am hoping to go into a pediatric specialty -- maybe genetics or endocrinology. I am really interested in working to bring specialized care to under-resourced communities. My time at the Clinic has shown me how a small group of dedicated people can radically change the healthcare landscape of a community for the better.

Joshua Wesalo After graduating in 2013, I worked for both the Clinic and my alma mater (F&M College) through an Eyler Fellowship. My

primary focus was researching GM3 Synthase Deficiency, a neurological disease that causes epilepsy and developmental problems in the Amish population. I was working on preparing GM3 and with the Clinic's UHPLC instrument to help streamline amino acid checks in patients with glutaric acidemia type 1 (GA-1) and to help measure patients' vitamin levels. I also had a few precious opportunities to shadow physicians when they saw patients. It was an honor to have the chance to work on the GM3 project and to meet the patients and families of these special children. This project reaffirmed that I wanted to combine scientific research in chemistry with medical practice in the future.

To that end, I am currently a student in the MD/PhD dual-degree program at the University of Pittsburgh. I am wrapping up my PhD in the laboratory of Dr. Alexander Deiters, where I am working on new research tools to control important proteins and nucleic acids using light and chemical triggers. I also work for an organization called Guerrilla Eye Service that provides free eye care to patients in need in the area. I plan to marry my research interests in chemistry with clinical interest in ophthalmology, and would like to split my time between running a lab, seeing patients, and teaching medical students in the future.

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2021 Clinic for Special Children 5k

Application Inside!

Join us on Saturday, September 18th from 9:00 a.m. - 11:00 a.m. for our annual Clinic for Special Children 5k! Details and application inside!

Adult Services at the Clinic

Read Katie Martin's story about her experience as an adult patient at the Clinic for Special Children. Learn about how we are expanding our services to provide care to patients as they transition out of traditional pediatric care.

Our 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."