



Clinic for  
Special Children

# Newsletter

ISSUE #49

SPRING 2020



## Treating Phenylketonuria (PKU) at the Clinic The Weaver Family

Estelle Weaver was born a week and a half early in August 2019. Her parents, Kenneth and Eve, were thrilled at the birth of their sixth child and second daughter. Before she left the hospital, Estelle received the state newborn “heel-stick” test. Four days later, they received the call that Estelle’s test was positive for Phenylketonuria (PKU). PKU is a genetic disorder that results in high levels of the amino acid phenylalanine. “The most shocking thing for us was that Estelle was our sixth child and we didn’t know that PKU ran in our family,” explains Kenneth.

The Weavers urgently called the Clinic for Special Children (CSC) and made an appointment the next morning with Dr. Vincent Carson. During that first three-hour long appointment, Dr. Carson explained what PKU was and discussed a treatment plan for Estelle’s care with her family. “We were so thankful that CSC was an option for us,” explains Eve. “We really appreciated the time that Dr. Carson took with us that first appointment and how he connected with us. It meant a lot to our family.”

Estelle’s treatment plan includes eating a low protein diet supplemented with a specialized formula that is phenylalanine free. Eve explains the transition for Estelle’s care, “It was a little bit of an adjustment to not breast feed Estelle, but every day mixing a doctor prescribed formula

recipe that is adjusted each week according to her levels was even more of an adjustment” Each week, she gets a heel prick test and sends it to CSC to monitor her phenylalanine levels. If a PKU patient is not treated, their phenylalanine levels can increase and cause irreversible brain injury resulting in intellectual disability and psychiatric issues.

Today, Estelle is a happy five-month old baby that loves playing with her siblings, especially when older sister Evelina sings to her in her rocking chair. To manage her PKU, Estelle will need to monitor her phenylalanine levels closely throughout her life. However, the future of PKU looks bright with the promise of new innovative therapies like gene replacement therapy. There are planned gene therapy trials for PKU and CSC is involved in ongoing research.

When asked about what message they would give to other parents, Kenneth and Eve say, “Allow yourself to grieve the loss of ‘normal’ for your child. Yet accept the new ‘normal’. Do what you can, the best you can for the future good of your child. Above all else remember to thank God for the roses and enjoy them, their beauty and fragrance, in spite of the thorns.”

Thank you to the Weaver family for sharing their story with Estelle and PKU!



## SPECIAL DATES

**2020 Auction Season  
June-September**

Find out more details inside!

**Rare Disease Day  
Saturday, February 29**

An international day to bring awareness to rare diseases around the world!

**Kumme Essa [Come Eat]  
A dinner to benefit the Clinic  
Thursday, April 2**

**Springside Barn**

Join us for Kumme Essa – our first dinner to benefit CSC! Tickets are required, additional details are inside on pages 2 and 5.

**Good Friday - Office Closed  
Friday, April 10**

All Day

**Memorial Day - Office Closed  
Monday, May 25**

All Day

**Translational Medicine Conference  
Thursday, July 30 - Friday, July 31**

**Franklin & Marshall College**

Join us for the 2020 Translational Medicine in Plain Populations Conference! See page 2 for more details.

**Clinic for Special Children 5k  
Saturday, September 19**

Mark your calendars and save the date for our 3<sup>rd</sup> annual 5k run/jog/walk!

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

## *Kumme Essa {Come Eat}*

*A Pennsylvania Dutch inspired dinner to benefit the Clinic*

**Thursday, April 2  
5:00 p.m. until 8:00 p.m.**

**at Springside Barn**

Join us for our first Kumme Essa dinner to benefit the Clinic for Special Children! Tickets are required to attend this dinner and are now available for sale (see page 5). The night includes hors d'oeuvres, a seated dinner, special talks, and a live auction!

For more information and details, please see page 5 of this newsletter. If you are interested in sponsoring the Kumme Essa dinner, please contact the CSC Development Team at 717-687-9407 or [eseitz@clinicforspecialchildren.org](mailto:eseitz@clinicforspecialchildren.org).

## *2020 Translational Medicine in the Plain Populations Conference*

**Thursday, July 30 – Friday, July 31  
All Day**

**at Franklin & Marshall College**

The Clinic for Special Children will be hosting the 2020 Translational Medicine Conference at Franklin & Marshall College in Lancaster, PA! This annual conference is largely organized by the Plain Community Health Consortium (PCHC), an association of clinics that share the mission of serving Old Order Amish and Mennonite (Plain) populations throughout North America.

The two-day conference focuses on clinical care and research of rare genetic conditions and will highlight novel research, innovative therapies, and thought-provoking concepts focusing on collaborative efforts between healthcare systems, academic partners, and PCHC. The agenda will include a keynote, common talks, and breakout sessions focusing on clinical, research, and administrative tracks.

The conference is open to the public and registration will begin in the coming weeks at [www.ClinicforSpecialChildren.org](http://www.ClinicforSpecialChildren.org) or by calling our office at 717-687-9407. Clinicians, scientists, midwives, administrators, patient families, rare disease advocates, and members of the Plain community are encouraged to attend. Advanced registration is required.

*Save the Date!*

**3<sup>rd</sup> Annual Clinic for Special Children  
5k Run/Jog/Walk**

**Saturday, September 19  
9:00 a.m. – 11:00 a.m.**

**at the Clinic for Special Children**

We are excited to announce our 3<sup>rd</sup> annual Clinic for Special Children 5k! Our 2019 race raised over \$40,000 and over 350 runners/joggers/walkers participated! Strollers and wheelchairs are welcome! Registration will open in late Spring/early Summer for the race– applications will be available at the Clinic, in the next newsletter, and on the Clinic's website.

If you are interested in becoming a sponsor for the 2020 Clinic for Special Children 5k, please contact the CSC Development Team at 717-687-9407 or [giving@clinicforspecialchildren.org](mailto:giving@clinicforspecialchildren.org).

*Save the Date!*

**Community Benefit Dinner  
Martindale Fellowship Center**

**Wednesday, October 14  
4:00 p.m. – 7:00 p.m.**

**at the Martindale Fellowship Center**

All are invited to join us for our 2020 Community Benefit Dinner at the Martindale Fellowship Center! Support and learn more about CSC with informational staff talks throughout the evening.

The dinner will be buffet-style with a variety of food and desserts! The dinner will be on a donation-only basis with the option of "eating in" or placing take-out orders.

We will have more details in the next newsletter regarding this exciting event!

## *2020 Benefit Auctions Website*

In addition to the included 2020 events calendar, you can now stay updated on our 2020 Benefit Auctions at [www.ClinicAuctions.org](http://www.ClinicAuctions.org)! The website is frequently updated throughout the benefit auction season with new information such as sale bills, flyers, photos, and more.

Information will also be updated on our home website ([www.ClinicforSpecialChildren.org](http://www.ClinicforSpecialChildren.org)) under 'Events' and on our Facebook page. We hope to see you at one of our 2020 benefit auctions!

## *Motor Assessment Study Looking for volunteers*

We are looking for healthy individuals (birth to 18 years old) to collect information on how healthy individuals perform three different motor assessments (measuring body movements). Motor assessments are used regularly to track progress and determine how well treatments are working for disorders such as spinal muscular atrophy (SMA). Collecting information on healthy children is essential to being able to interpret results in children with muscle issues.

The assessments will take about an hour at Strasburg Mennonite Church. If interested in helping with this study, please call Lauren at the Clinic at 717-687-9407. We appreciate your help!

## *2020 Family Day Schedule*

Each year the Clinic hosts a variety of family days to update patients and their families on disease-specific updates, new treatments, and best practices. The days feature talks from CSC staff members and collaborators and also allow patient families to have a day of fellowship with others who are experiencing similar challenges. Invitations will be sent out to applicable families for the various family days.

The below family days are currently scheduled for 2020 through July. Stay tuned for the rest of our 2020 family days in upcoming newsletter announcements:

**Bereavement Day  
Midwife Conference  
Mental Health & Wellness Day  
Special Education Family Day**

## *CSC Web Shop*

Have you visited CSC's online shop yet? We have a variety of items for sale including mugs, notecards, and books! You can purchase your copy of CSC's newest books, *30 Stories for 30 Years* and *Our Story in Newsletters*.

To visit our online shop, go to [www.ClinicforSpecialChildren.org/store](http://www.ClinicforSpecialChildren.org/store). You can also purchase any items from our web shop in-person at CSC. All proceeds from the sales on our web shop benefit the Clinic for Special Children and directly support our mission.





## Mark your Calendars for the 2020 Benefit Auctions!

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

### 2020 Auction Season Kick Off!

You're invited to join us during our 2020 Auction Season! This year we have six benefit auctions throughout Pennsylvania, Ohio, and Missouri! Each auction features a day of handmade and unique items for sale, delicious food, fellowship, and more!

The six benefit auctions make up about 30% of the Clinic's annual operating budget, which allows us to continue providing life-saving care to those with rare genetic disorders.

Please visit our auctions website ([www.ClinicAuctions.org](http://www.ClinicAuctions.org)) to view the most current auction information for the 2020 season.

#### Union County Auction | 6.6.20

Flower Sale and Rib Dinner | 6.5.20 | 5-8 p.m.

Buffalo Valley Produce Auction

22 Violet Road

Mifflinburg, PA 17844

**Contact:** Leon Hoover | 570-966-2414

#### Lancaster County Auction | 6.20.20

Leola Produce Auction

135 Brethren Church Road

Leola, PA 17540

**Contact:** Mark Martin | 717-733-3070

*\*Breakfast starts at 6:30 a.m.*

#### Shippensburg Auction | 6.27.20

Cumberland Valley Produce Auction

101 Springfield Road

Shippensburg, PA 17257

**Contact:** Elvin Oberholtzer | 717-532-9088

#### Ohio Auction | 7.11.20

Blooming Grove Auction Inc.

1091 Free Road

Shiloh, OH 44878

**Contact:** Michael Newswanger | 419-896-2184

#### Missouri Auction | 8.15.20

Ed Good Family Farm

10507 County Road 813

Memphis, MO 63555

**Contact:** Harlan Burkholder | 660-341-4113

#### Blair County Auction | 9.12.20

Morrison's Cove Produce

4826 Woodbury Pike

Roaring Spring, PA 16673

**Contact:** Mervin Martin | 814-793-3529



**"It's important to recognize how far we've come and to celebrate the successes. But it's MORE important to remain humble and remember how much more work we have to do."**

*-Dr. Vincent Carson, Pediatric Neurologist*





## A Year in Review 2019 at the Clinic for Special Children

The Clinic for Special Children had an exciting and busy 2019! We are so grateful for the support and are excited for what 2020 has in store!

### 2019 BENEFIT AUCTION SEASON

The 2019 benefit auction season was a record-breaking one! Across all six (6) auctions, over \$1 million was raised to support the Clinic's mission – making up approximately a third of our operating budget. Several individual auctions also broke their fundraising records. Join us for a benefit auction in 2020 for amazing days of fellowship, great food, unique items for bid, and more!

### 30<sup>th</sup> ANNIVERSARY

The Clinic celebrated its 30<sup>th</sup> anniversary throughout 2019. We held our 30<sup>th</sup> anniversary celebration event in April and welcomed patient families, donors, collaborators, and supporters to the Clinic and featured "Taste of Lancaster County" food. We also published two books to mark the occasion, "30 Stories for 30 Years" and "Our Story in Newsletters: 1989–2018". In addition, we had a commemorative fraktur created by local artist, Lynn Sommer, and had a special 30<sup>th</sup> anniversary mug for sale.

### SMA PREVENTION READINESS

The SMA Prevention Readiness program was a great success in 2019. Since Spinal Muscular Atrophy (SMA) is now treatable but early diagnosis is key, this program provided free carrier testing for adults. Originally planning to collect 2,000 samples over 3 years, CSC has conducted over 2,177 SMA carrier tests in 15 months. The program identified or confirmed 318 individual carriers and nine (9) carrier couples, where both spouses are carriers. This allowed for three (3) affected infants to be identified and treated with gene therapy presymptomatically. Another six (6) infants were tested as newborns due to having carrier parents and

were not affected. We'd like to thank everyone that's participated or volunteered in the SMA Prevention Readiness program!

### PLAIN INSIGHT PANEL™

In the Summer of 2019, the Clinic launched the Plain Insight Panel™, a next generation sequencing (NGS) panel that allows us to test for over 1,300 genetic variants in a single test. The partnership with Nemours Alfred I. duPont Hospital for Children, ArcherDX, and the Plain Community Health Consortium (PCHC) made this project possible. To date, we've conducted carrier testing through the Plain Insight Panel™ for over 400 individuals. Funding from the Stabler and Anabaptist Foundations allowed us to offer this \$500 test at \$99 for carrier testing.

### 2019 CLINIC FOR SPECIAL CHILDREN 5k

Our 2<sup>nd</sup> annual Clinic for Special Children 5k was held in September of 2019. Over \$40,000 was raised for the Clinic with over 350 registrants, a bake sale, silent auction, and our first free kid's fun run. Thank you to all of the participants, corporate sponsors, volunteers, and everyone who made the day possible! Save the date for our 3<sup>rd</sup> annual Clinic for Special Children 5k on Saturday, September 19, 2020.

### GENE THERAPY

The Clinic serves as a clinical trial site for the Sprint trial evaluating AVXS-101 in pre-symptomatic patients with SMA Types 1, 2, and 3. Dr. Kevin A. Strauss has traveled the world this past year to present key findings from the trial. In 2020, the Clinic will be serving as a clinical trial site for up to four separate gene therapy trials, a commitment to translational research that places the Clinic alongside internationally recognized academic universities and hospitals, and ensures that our patients have access to cutting-edge therapies.



# RECENTLY PUBLISHED PAPERS

**Kevin A. Strauss, Vincent J. Carson,** Kyle Soltys, **Millie E. Young, Lauren E. Bowser, Erik G. Puffenberger, Karlla W. Brigatti, Katie B. Williams, Donna L. Robinson, Christine Hendrickson, Keturah Beiler,** Cora M. Taylor, Barbara Haas-Givler, Stephanie Chopko, Jennifer Hailey, Emilie R. Muelly, Diana A. Shellmer, Zachary Radcliff, **Ashlin Rodrigues, KaLynn Loeven, Adam D. Heaps,** George V. Mazariegos, D. Holmes Morton. **Branched-chain  $\alpha$ -ketoacid dehydrogenase deficiency (maple syrup urine disease): Treatment, biomarkers, and outcomes.** *Molecular Genetics and Metabolism.* 16 January 2020. doi: 10.1016/j.ymgme.2020.01.006. [Epub ahead of print]

Kang SK, Vanoye CG, Misra SN, Echevarria DM, Calhoun JD, O'Connor JB, Fabre KL, McKnight D, Demmer L, Goldenberg P, Grote LE, Thiffault I, Saunders C, **Strauss KA,** Torkamani A, van der Smagt J, van Gassen K, Carson RP, Diaz J, Leon E, Jacher JE, Hannibal MC, Litwin J, Friedman NR, Schreiber A, Lynch B, Poduri A, Marsh ED, Goldberg, EM, Millichap JJ, George AL Jr, Kearney JA. **Spectrum of Kv 2.1 Dysfunction in KCNB1-Associated Neurodevelopmental Disorders.** *Annals of Neurology.* 2019 Dec;86(6):899–912. doi: 10.1002/ana.25607. Epub 2019 Oct 24

Iffland PH 2nd, **Carson V,** Bordey A, **Crino PB. GATORopathies: The role of amino acid regulatory gene mutations in epilepsy and cortical malformations.** *Epilepsia.* 2019 Oct 17. doi: 10.1111/epi.16370. [Epub ahead of print]

\*bolded names indicate CSC staff or board authors

## Kumme Essa {Come Eat}

A Pennsylvania Dutch inspired dinner to benefit the Clinic for Special Children

Join us for our first dinner to benefit the Clinic for Special Children at Springside Barn in East Earl, PA. Tickets are limited, please purchase yours today!

Enjoy an evening with CSC staff, collaborators, and supporters for a family-style meal, live auction, hors d'oeuvres, and more!

### Thursday, April 2, 2020

5:00 p.m. until 8:00 p.m.

**Springside Barn**

1294 Weaverland Road, East Earl, PA 17519



**Tickets** \$150 *per ticket*

*For parties of 8 or more, please contact us regarding special pricing.*

**To reserve tickets** (reply requested by Thursday, March 19<sup>th</sup>):

**Purchase Online**  
at [ClinicforSpecialChildren.org](http://ClinicforSpecialChildren.org)

**Mail a check\***  
Clinic for Special Children  
Attn: Development Team  
535 Bunker Hill Road  
Strasburg, PA 17579

**Contact Us**  
717-687-9407  
or  
[rsvp@ClinicforSpecialChildren.org](mailto:rsvp@ClinicforSpecialChildren.org)

*\*If mailing a check, please include a list of guest names and note dietary restrictions*

## Sponsorships

Sponsorship packages are available for the Kumme Essa dinner. If you are interested in reserving a sponsorship package, please contact our Development Team at 717-687-9407 or [eseitz@clinicforspecialchildren.org](mailto:eseitz@clinicforspecialchildren.org).





Not pictured: Lavina King, KaLynn Loeven, Julia Martin, Stephen Ratcliffe, MD, MSPH, Millie Young

## Local and Regional Foundations Support the Clinic's Growth

*Written by Emily Seitz, Development Director*

The Clinic for Special Children has grown tremendously in the past several years, from adding physicians to care for our patients aging out of pediatric care and a genetic counselor to meet the increasing needs of our patients and their families interested in their genetic health, as well as expanding *Cherished Lives*, our palliative care program for patients living with life-limiting illnesses. As we work to expand and better serve the needs of our patients, their families, and the community, we have been incredibly fortunate to receive support from local and regional foundations to kick-start our work in this formative period.

Growth requires that we secure our infrastructure, and a \$25,000 matching grant from The Steinman Foundation's Community Stewardship program makes possible the Clinic's transition from our current electronic medical record (EMR) system to Epic – a cutting-edge, universal, and secure system offered in partnership with Penn Medicine Lancaster General Health's (LGH) Community Connect program. This transition is critical to the Clinic's ability to provide the highest quality personalized genomic medical care to our nearly 1,100 active patients. Specifically, the benefits of transitioning to the Epic system are fourfold: first, the new system will interface seamlessly with the Clinic's primary medical collaborators to ensure continuity of care for our patients. Second, this transition ensures the security and integrity of our patient records. Third, transitioning to Epic allows our patients increased access to their medical information on a level the Clinic is presently unable to provide. Finally, the partnership with LGH makes the conversion to Epic a financial possibility that the Clinic otherwise would be unable to afford on its own, and represents a savings of nearly \$100,000.

Attending to our infrastructure frees the Clinic's staff to do the work that really matters: creating innovative programs like the Plain Insight Panel™ (PIP) that better serve the needs of our patients and communities. In 2018 our researchers and clinicians worked with the local Plain communities and scientific and industry partners to develop the PIP, a genetic test for individuals of Plain descent used to determine their risk for bearing children with genetic disorders found in these communities, as well as personal implications for health and wellbeing. This test represents the culmination of over thirty years of effort on the part of Clinic researchers and clinicians

to identify and treat genetic disorders common in the Plain communities of Central Pennsylvania, enabling rapid diagnosis and personalized clinical management – a central tenant of the Clinic's mission. The Donald B. and Dorothy L. Stabler Foundation and the Anabaptist Foundation both supported the early phases of this program and allowed the Clinic's staff to simultaneously offer the test at the discounted rate of \$99 per person and begin analyzing patient data to refine the clinical process and publish the results so that the study's findings can be broadly communicated to the scientific and clinical communities. Most importantly, funding from the Stabler and Anabaptist Foundations allowed parents to use the results of the panel to provide the best possible care for their newborn child while avoiding unnecessary medical costs.

Alongside creating the PIP, the Clinic's physicians and nursing staff are similarly invested in caring for our patients with life-limiting genetic illnesses for which no treatment is currently available. Our *Cherished Lives* program provides culturally-sensitive comfort care and grief and counseling resources for Plain patients and their loved ones. A recent \$10,000 gift from Wellspan Health's Community Partnership Grants Program allows us to formalize this vital service through providing memory gifts, grief resources, and medical equipment to our Plain patients and their families for no cost. And because community is a vital part of the grieving process, this gift will also enable the Clinic's clinicians to host a bereavement family day for our patient families so that they can learn more about helpful resources available to them at a particularly difficult time, as well as seek support amongst other families who may be on the same journey. To be sure our clinicians have access to the most up-to-date palliative practices, this gift also provides professional development funds for Keturah Beiler, the leader of this impactful nurse-led program.

On behalf of our patients and their families, the Clinic's staff is deeply grateful to the Steinman Foundation, the Anabaptist Foundation, the Donald B. and Dorothy L. Stabler Foundation, and Wellspan Health's Community Partnership Grants Program for their support of the early stages of these vital projects – support that places us on strong footing to continue creating innovative programs to better serve our patients and their families.





## Collaborator Spotlight: Dr. Aaron Chidekel, Pulmonology, Nemours Children's Health System

*Written by Dr. Chidekel*

A pediatric pulmonologist is a physician who cares for children of all ages with lung and breathing problems. We are trained and certified as pediatricians first, and then spend an additional three years learning our subspecialty. Some pediatric pulmonologists also have additional expertise in sleep medicine and I have this certification as well. What this means is that I am trained to recognize, diagnose and treat acute and chronic lung and breathing problems of all types. An example of an acute condition would be a severe pneumonia and the most common chronic lung disorder that pediatric pulmonologists treat in children is asthma.

While I have been living in southern Chester County, PA and working at Nemours in Wilmington, DE for almost 25 years, practicing and raising my family, I am originally from the New York metropolitan area. I am neither a Rangers nor a Giants fan however! My education and training occurred in New England. I went to college and medical school at Brown University and did my Pediatric Residency and Post-Doctoral Fellowship in Respiratory and Sleep Medicine at Yale-New Haven Hospital and Yale University respectively. After completing all of this training, I moved to southern Chester County with my wife Stacy to start working at Nemours and have happily been there ever since. This has been a wonderful place to grow my career and raise our three children.

I became interested in pulmonary medicine during my Pediatric residency after learning about Cystic Fibrosis, and this genetic cause of lung disease remains a passion. However, many other genetic diseases affect the lungs and breathing mechanism and therefore, a large part of my practice involves the management of children with other types of inherited, or genetic diseases. Examples of these diseases that I see at the Clinic for Special Children (CSC) include congenital muscular dystrophies, skeletal dysplasia syndromes such as Ellis-Van Creveld Syndrome (EVC) and Trisomy 21 or Down Syndrome. Children with immune deficiencies also commonly develop lung disease as well.

Sleep disordered breathing and other sleep problems are a large part of my practice now as well. Over the past 10-15 years, we have learned that obstructive sleep apnea is much more common in children than previously thought, and unfortunately, with the epidemic of obesity in America we are treating many children with sleep-disordered breathing related to unhealthy weight. Other sleep disorders that we see also include common behavioral sleep problems, nightmares or night terrors and even much rarer conditions such as narcolepsy. Yes, we do see

narcolepsy in children!

I was actually introduced to CSC by Dr. O'Reilly who is one of the Ears, Nose and Throat (ENT) physicians who previously worked at Nemours. He invited me out to the Clinic, introduced me to Dr. Strauss and the rest is history as they say. That was probably 4 or 5 years ago now, but it is hard to remember as time goes by quickly when the work is rewarding and enjoyable. At CSC, I collaborate very closely with everyone on the team, but due to the particular impact of congenital muscle and cardiac diseases on the lungs and breathing, I share many of my patients with Dr. Carson at CSC and Dr. Chowdhury at Cardiology Care for Children. I care for children with many other types of respiratory conditions as well. Some simply have asthma, but this is not very common in the Amish Community. Some children need supplemental oxygen, tracheostomy tubes or other types of technology-dependence and some are simply recovering from a severe respiratory tract infection.

When I was in Medical School, I spent three months in Shiprock, New Mexico on the Navajo Reservation. This clinical rotation with the Indian Health Service remains one of the more meaningful "medical" experiences I have had in my career. In fact, my wife Stacy who trained as an Art Historian, and I were even considering going out West for a period of time after my training to work in the Navajo community. There are many parallels to this experience that make my time and work at CSC so meaningful as well. Seeing challenging medical cases in a uniquely special cultural context, and integrating these factors into the family-centered care of an often chronically ill child is incredibly meaningful. This is the care that is delivered at CSC every day, and I am privileged to be a small part of the team.

I am usually at CSC at least once per month. I will often start or end my day with a house call to see a technology-dependent patient for whom travel to CSC is particularly difficult. As I mentioned already, I will often see patients alongside Dr. Chowdhury, as we share many of the EVC patients or children with Trisomy 21 and co-existent heart and lung disease, or with Dr. Carson as we share some of the muscular dystrophy patients. I also see patients from CSC when they are hospitalized at Nemours in Delaware and can help with the continuity of care in this fashion.

Seeing children and families at the Clinic for Special Children has become a "special" part of my overall practice. I am grateful to be included in this important, community-focused holistic work and look forward to continuing to contribute to the patients and care team at CSC for many more years.



## Our Staff

Keturah Beiler, RN  
Nurse

Lauren Bowser  
Research Fellow

Karlla Brigatti, MS, LCGC  
Research Operations Director

Vincent Carson, MD  
Pediatric Neurologist

Kelly Cullen  
Communications Manager

Adam D. Heaps, MS, MBA  
Executive Director

Christine Hendrickson, RNC  
Nurse

Candace Kendig  
Medical Receptionist

Lavina King  
Community Liaison

Yalonda L. Kosek  
Office Coordinator

KaLynn Loeven  
Laboratory Scientist

Julia Martin  
Development Assistant

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, LCGC  
Genetic Counselor

Emily Seitz  
Development Director

Kevin A. Strauss, MD  
Medical Director

William Van Ess, MS, CFE  
Accountant

Millie Young, RNC  
Research Nurse

## Board of Directors

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Stephen Tift, MD

Glen Zimmerman  
Chair-Development Committee

Jacob Zook  
Vice Chairman



## Donor Spotlight

### *The Promise Players*

In the early 1990's, Doreen Creighton participated in a one-day acting seminar organized by a retired minister in Parkesburg, PA. That minister put on a short play in which she participated, unexpectedly caught the acting bug, and went on to act in a few skits and plays at her church, Lampeter Church of the Brethren. In those early acting years, she explains how the preparation impacted her, "It struck me how as an actor you assume that you're preparing for your audience, but the play ends up impacting you as an actor just as profoundly." Doreen then had the idea of developing a type of "community theater" received the approval from her pastor, and The Promise Players was born.

The Promise Players is a non-profit organization comprised of a group of volunteer community actors and committee members. Since their first play in the Fall of 1995, "Pokeweed and Mrs. Gasp and Other One-Acts With a Point-of-View", they have performed 48 plays and 374 performances. Over the years they have put on a variety of shows including historical dramas, a play similar to the "Left Behind" series, and comedy. In 2020, they will be celebrating their 25<sup>th</sup> season.

Comedy is a theme that they keep coming back to as "comedy and humor can get the message across in such an easy way." The journey of The Promise Players is a unique one, with Doreen explaining "We are an organization that should not exist. I never did acting in high school, college or had formal training, other than a few local workshops. I had an interest, went to the retired minister, and took the one-day seminar. It started with nothing, but through God it's become such a blessing to everyone involved and a humbling experience."

From the beginning, The Promise Players choose a charity to receive all proceeds from their plays. In total, they have raised \$238,700 for over 40 national, regional, and local charities. In November of 2019, The Promise Players chose the Clinic for Special Children to benefit from their "Don't Kick the Turkeys" play. It was their biggest grossing show yet - resulting in a donation of over \$14,000 to support the Clinic's mission!

In May of 2020, The Promise Players will be featuring "The Case of the Duplicate Date Nut Loaf" - their first dinner theater play! It will also be the first fundraising play to benefit The Promise Players to help update their technology systems such as lighting and microphones. They also plan to do "Smoke on the Mountain" on March 20<sup>th</sup>-22<sup>nd</sup> and 26<sup>th</sup>-29<sup>th</sup>, 2020. The Promise Players accept donations directly to help support their operations. We are so grateful to The Promise Players for their amazing support of the Clinic for Special Children!





# WHAT'S GOING ON AT THE CLINIC?



Dr. Kevin Strauss and Karlla Brigatti, MS, LCGC are pictured in a group meeting in São Paulo, Brazil at the MSUD/GA1 Summit at the National Neurology Congress.



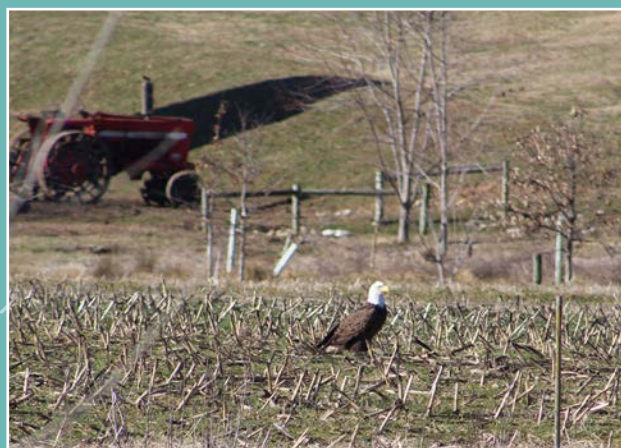
Dr. Kevin Strauss delivers a talk about the Clinic for Special Children at the SBNI Congress in Brazil.



Dr. Vincent Carson delivers a talk at the Woodcrest Villa Retirement Community in Lancaster, PA about the Clinic for Special Children and the history of research at the Clinic.



CSC team members in downtown Lancaster during the 2019 ExtraGive! The Clinic raised over \$77,000 in 24 hours and surpassed our goal of \$70,000!



We spotted a special visitor in our fields in January!



Dr. Kevin A. Strauss delivered a talk at the "Beyond a Million Genomes: From Discovery to Precision Health" conference in Breckenridge, CO.





## Clinic for Special Children

535 Bunker Hill Road  
PO Box 128  
Strasburg, PA 17579

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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.

# 2020 Benefit Auctions for the Clinic for Special Children

See inside for more details!

## June

**6** Union County, PA

**20** Lancaster County, PA

**27** Shippensburg, PA

## July

**11** Shiloh, OH

## August

**15** Memphis, MO

## September

**12** Blair County, PA

### The Clinic's Mission

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

Charles H. Winters, Jr.  
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