Clinic *for* Special Children®

Newsletter

ISSUE #57 FALL 2022

Life with Ellis-van Creveld Syndrome
The Stoltzfus Family

"I knew within 30 seconds of Becky being born that she had EVC (Ellis-van Creveld Syndrome)," remembers Linda Stoltzfus. Just hours after Linda and her husband, Abner, welcomed Becky into the world, they were surprised to find themselves at the Clinic for Special Children. "Our midwife stressed to us that we needed to get Becky's heart checked out immediately," says Linda. After assessing Becky's condition, the Clinic decided that she needed urgent complex care and sent her to Penn Medicine Lancaster General Health's Women's and Babies Hospital. To learn more about EVC, visit page 4 of this newsletter.

At the hospital, the family met Dr. Devyani Chowdhury, the director of Cardiology Care for Children, who serves as a visiting pediatric specialist at the Clinic. Dr. Chowdhury recommended cardiac intervention and an echocardiogram showed that Becky required life-saving surgery or she would not survive. Becky was then flown to Nemours Children's Hospital in Wilmington, Delaware. Becky was initially at Nemours for a three-month stay, which included a hybrid procedure as she was too sick to have open heart surgery and there were many difficulties in stabilizing her condition. "It was once she got a procedure to fix the backup of stomach contents into her esophagus (Nissen fundoplication) with a G-tube (feeding tube) around three months old that she really started to turn around and we were able to bring her home," explains Linda and Abner.

After her discharge from Nemours, Becky was connected with Dr. Kevin Strauss, Medical Director at the Clinic, who saw Becky at her first visit and has been her primary care provider ever since. Becky was also connected to multiple visiting specialists at the Clinic for her comprehensive care.

"Around her first birthday, Dr. Chowdhury started to mention about going to Boston Children's Hospital to get the intensive heart surgery that Becky needed. Shortly after we were on our way to Boston," says Linda. "At first we were not impressed about going so far, but we've never been sorry for they helped her so much."

At Becky's first visit to Boston Children's Hospital, she had catheterization procedures to optimize her cardiac plumbing for a two chamber heart repair surgery. A year later when she was older and healthier for surgery, they went back to Boston Children's Hospital where Becky had a two chamber surgical intervention. To this day, it's thankfully the last heart surgery she's needed.

"Over the first two years of Becky's life, we were at the Clinic frequently and also saw Dr. Chowdhury as a visiting specialist at the Clinic," says Abner and Linda. "To have visiting specialists on-site at the Clinic is so valuable. It would have been so much further and more expensive for us to have to travel to see all of Becky's specialists, but instead, they came to the Clinic."

"When we were at Nemours for the first three months of Becky's life, we didn't even realize that the Clinic was following our story the whole time. I remember coming to the Clinic the first visit after getting discharged from the hospital and everyone was cheering us on when we walked in," remembers Abner and Linda. Becky has also had several non-cardiac surgeries, all coordinated through the Clinic with support from cardiology.

Today, Becky is a happy and healthy eight-year-old. She is a great helper around the house, especially with her younger siblings. "To have an EVC baby, it's not always an easy road but it's also not a big deal," notes Linda, "we trusted whatever the Clinic and Dr. Chowdhury told us to do and they were there for us along the journey."

SPECIAL DATES

Finger Lakes Benefit Auction *NEW*
Saturday, October 1
Ontario Produce Auction | Stanley, NY

Community Benefit Dinner
Wednesday, October 12
Martindale Fellowship Center
4:00 p.m. - 7:30 p.m.
A seafood and ham style buffet dinner to benefit the Clinic! Details inside.

The Extraordinary Give
Friday, November 18
ExtraGive.org
Online Fundraiser - All Day
Support the Clinic during this 24-hour giving day! Details inside.

Thanksgiving | Office Closed
November 24 & 25
All Day

Christmas Eve | Office Closed
Friday, December 23
From 1 p.m. - 5 p.m.

Christmas Holiday | Office Closed
December 26 & 27
All Day

New Years Eve | Office Closed
Friday, December 30
From 1 p.m. - 5 p.m.

New Years Day | Office Closed
Monday, January 2
All Day

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

Updates from Clinic for Special Children®

Staff News

Candace Kendig, RMA

Congratulations to Candace, Office Manager at the Clinic, for celebrating her five year work anniversary!

Candace joined our team on July 3, 2017 as a Medical Receptionist. Candace works with our clinical and administrative teams and oversees our front desk operations, patient assistance programs, and more! She recently earned her bachelor's degree in healthcare management from Central Penn College.

KaLynn Loeven

Congratulations to KaLynn, Laboratory Scientist at the Clinic, for celebrating her five year work anniversary!

KaLynn joined our team on July 10, 2017 as a Laboratory Technician. She works everyday in our lab at the Clinic and is responsible for performing genetic research through microarrays and developing new testing assays.

Kelly Milligan

We're excited to welcome Kelly to the Clinic! Kelly joined the staff in June as the organization's first Event and Volunteer Manager.

In this role, Kelly will focus on building and stewarding an organization-wide volunteer program and serving as the primary manager of Clinic-sponsored events. She will also be involved in event coordination for organization-wide events like disease specific family days and employee events.

In her free time, Kelly loves to garden, spend time outdoors, socialize with friends and family, and assist people in planning their weddings.

Welcome to the Clinic, Kelly!

Ashton Bollinger

We're excited to welcome another new member to our team, Ashton! Ashton joined the staff in June as a Research Associate. She succeeded Emilienne Bolettieri in the role, as Emi headed to medical school this summer.

In this role, Ashton will support a number of the Clinic's research endeavors. These projects will provide insights into rare genetic conditions and she will be involved with prospective history studies, clinical trials of gene replacement therapies, and the analysis of vital data.

In her spare time, Ashton enjoys spending time outdoors and exploring nature, as well as stargazing on nights when the sky is clear. She also has a lot of fun with cooking and baking!

Welcome to the Clinic, Ashton!

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 six months old or older. Please contact your doctor with questions regarding the COVID-19 vaccine. If you are interested in receiving the COVID-19 vaccine, 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.

Office Operations

We have updated our mask guidance for staff and visitors to our clinic facility. Since community level COVID transmission is low in Lancaster, masks are currently optional for staff members and visitors who are asymptomatic. If staff or any visitors to the Clinic have any symptoms of illness or a known COVID exposure, they will be asked to mask in our facility. Going forward, we will continue to monitor the community level of COVID transmission and will adjust our guidance as necessary.

We are continuing to ask that if you have a scheduled appointment and are feeling unwell, please give us a call to let us know before your appointment at 717-687-9407. If you have any questions before your visit, please contact us at 717-687-9407.

Join us!
Community Benefit Dinner

Wednesday, October 12th
4:00 p.m. - 7:30 p.m.
at the Martindale Fellowship Center

The annual Community Benefit Dinner is slated for Wednesday, October 12th from 4:00 p.m. until 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.

We hope to see you there for a fun night of delicious food and great fellowship all in support of the Clinic!

It's Flu Shot Season

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 an annual flu shot.

We offer flu shots to our patients and their immediate family members for $20 per person. The best way to protect yourself and your family is to avoid exposure, practice good handwashing, and receive an annual flu shot.

EXTRA GIVE™

Lancaster County Community Foundation · Rodgers & Associates Wealth Advisers · High Foundation

You Can Help Make an *Extraordinary Difference!*

We're counting down the days until the **Extraordinary Give** on **Friday, November 18th** - Lancaster County's largest day of online giving! Help us **raise $100,000 in 24 hours** for the Clinic's mission! Last year, Clinic supporters raised over $110,000 in 24 hours! Every dollar you donate on November 18th during the ExtraGive will be stretched by a pool of more than $500,000 from local sponsors.

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

1. On Friday, November 18th (from midnight to 11:59 p.m.) **visit the website ExtraGive.org** and select 'Clinic for Special Children' to give online during the 24-hour period.
2. **Call the Clinic at 717-687-9407** between 9:00 a.m. - 5:00 p.m. to donate via credit card over the phone. Won't have access to internet on the day of ExtraGive? Call us before or on November 18th, and we'll help process your ExtraGive donation for you on the day of ExtraGive.
3. **Set up your own fundraising page!** On the day of ExtraGive, share your page with family and friends to raise money in support of the Clinic for Special Children! Visit ExtraGive.org, search for 'Clinic for Special Children', and click the teal 'Champion' button to create your own page before November 18th!
4. On Friday, November 18th visit our info table at Speckled Hen in Strasburg from 10:00 AM until 2:00 PM! Enjoy a delicious cup of coffee and support the Clinic during the ExtraGive!

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

RECENTLY PUBLISHED PAPERS

***Kevin A. Strauss**, Michelle A. Farrar, Francesco Muntoni, Kayoko Saito, Jerry R. Mendell, Laurent Servais, Hugh J. McMillan, Richard S. Finkel, Kathryn J. Swoboda, Jennifer M. Kwon, Craig M. Zaidman, Claudia A. Chiriboga, Susan T. Iannaccone, Jena M. Krueger, Julie A. Parsons, Perry B. Shieh, Sarah Kavanagh, Sitra Tauscher-Wisniewski, Bryan E. McGill, and Thomas A. Macek.* **Onasemnogene abeparvovec for presymptomatic infants with two copies of *SMN2* at risk for spinal muscular atrophy type 1: the Phase III SPR1NT trial.** Nature Medicine. 2022 Jun 17. doi: 10.1038/s41591-022-01866-4. Online ahead of print.

***Kevin A. Strauss**, Michelle A. Farrar, Francesco Muntoni, Kayoko Saito, Jerry R. Mendell, Laurent Servais, Hugh J. McMillan, Richard S. Finkel, Kathryn J. Swoboda, Jennifer M. Kwon, Craig M. Zaidman, Claudia A. Chiriboga, Susan T. Iannaccone, Jena M. Krueger, Julie A. Parsons, Perry B. Shieh, Sarah Kavanagh, Melissa Wigderson, Sitra Tauscher-Wisniewski, Bryan E. McGill, and Thomas A. Macek.* **Onasemnogene abeparvovec for presymptomatic infants with three copies of *SMN2* at risk for spinal muscular atrophy: the Phase III SPR1NT trial.** Nature Medicine. 2022 Jun 17. doi: 10.1038/s41591-022-01867-3. Online ahead of print.

**bolded names indicate current CSC staff authors*

Living with Ellis-van Creveld Syndrome

An interview with CSC's Community Liaison, Lavina King

As a close neighbor to the Clinic for Special Children, a member of the Old Order Amish community, and a person living with a rare genetic disorder, Lavina King knows all too well the impact that the Clinic has had on many families over the years. After learning about the Clinic's mission several years ago, Lavina joined the Clinic staff part-time as a Community Liaison. Lavina has lived with Ellis-van Creveld Syndrome (EVC) for decades, and shared her experiences with us through an interview.

What was your early childhood like living with EVC?

My parents knew right away when I was born that I had EVC. However, I didn't realize that something was really different with me until I started school and started riding the bus. Naturally, my mother was worried about me keeping up and being able to do things that the other kids would do. I was very bashful as a child and my sister, who also has EVC, didn't go to the same school as me. For the most part, the teachers and other students were very good and kind and accepting of who I was - which was a huge blessing.

What are some challenges that you've faced and how have you dealt with them?

My sister and I had healthy years growing up, which we are very thankful for as many EVC children have heart issues and/or orthopedic issues. We didn't have the Clinic for Special Children back then, and it would have been such a blessing to have at the time.

While I had great school years, and it wasn't until I grew older, about 16 years old, that it was a harder time for me to face and a huge milestone of my life. It was an intimidating time to start going out with the youth, however, most people were accepting and I ended up meeting my husband, Phares, and had a son named Leroy (who is now a grown young man!). After I had Leroy, I ended up getting heart surgery and in more recent years, have had some challenging orthopedic surgeries.

Overall, I really didn't think much about having EVC growing up. It wasn't until I was older that I came to accept it very well, got married, and had a child - a normal life like most other people. At times when I go into a crowd of people I can hear children or people say things, but it impacted me a lot more as a child. Now when I see someone staring in my direction, I'll look back at them and give them a smile.

What advice would you give to families with children with EVC?

For parents, I would say to just accept your children as they are as much as possible and to not make a big deal out of it. It's fine to talk to your children about what they have and what they are facing, but always treat them like everyone else. You can show your child that they can do anything that they put their mind to. The feelings that they experience are real when someone looks at them as different or looks down on them. Listen to your children when they share their feelings and know that what they are feeling is real to them.

In my role as a Community Liaison at the Clinic, I feel blessed to reach out to the younger generation and feel led to help and support the next generation of children with EVC. Honestly, I'm drawn to little people like a magnet.

What is a common misunderstanding that people have about EVC or rare genetic disorders in general?

Often people would look at others with a rare genetic disorder, or in a wheelchair, and would not realize that the person is just like us. I've learned so much myself by working at the Clinic that many of our patients may have physical disabilities that do not affect their mental state. They're just like everyone else and want you to talk to them as a person or adult.

After speaking with one mom whose daughter goes to the Clinic, she mentioned that her daughter does not appreciate when people walk in the room and start talking to her like she's a toddler. She's an adult and just wants to have a conversation just like any other adult. Overall, say hello to everyone you meet and accept them for who they are.

What do you wish for the future? Specifically for EVC?

One of my life lessons has been to be content and accept where God plants you in life. I know some people with EVC are married and some are not, but be content with God's life plan for you. I always think to myself about how I never thought that I'd get married and have a family of my own when I was growing up. I actually had dreamed of a life on my own and having my own house until I met my husband Phares.

For the medical side of EVC, I've already seen so much progress in my lifetime with the heart surgeries that are performed now and the orthopedic help that patients get. I would say for the future of taking care of EVC patients, to keep them as healthy as possible and to be there for them. Even if there is the possibility to have therapies that can reduce suffering for more severe cases of EVC, it would be a blessing.

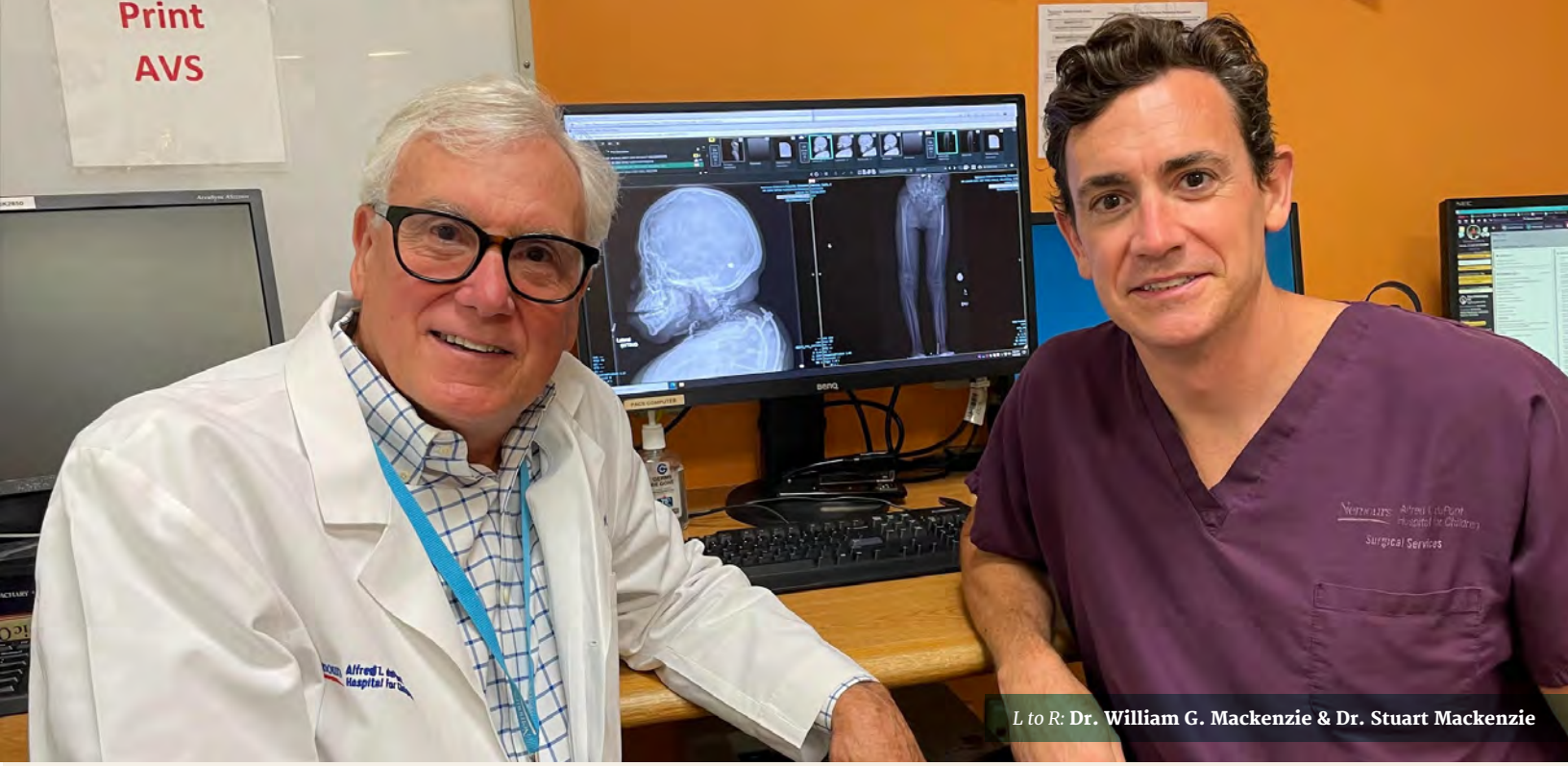
The Clinic has done so much for EVC patients over the years. Especially with connecting families to speciality care on-site like cardiology services or orthopedic surgery (see page 5 for a collaborator spotlight of orthopedic surgeons that visit the Clinic).

I also think that continuing to connect EVC families together so that they can support each other. It's huge for young adults with EVC to have friends that also have EVC - it can be such a therapeutic relationship to have. I'm part of a group of women with EVC and we have such a strong bond. Even though we live in different areas, we get together once in a while and will write to each other consistently. Even if it's at a distance, keep connecting with others that are facing the same things that you are.

What is Ellis-van Creveld Syndrome?

Ellis-van Creveld Syndrome (EVC) is a rare genetic condition that affects development of the heart, lungs, ribs, limb bones, nails, and teeth. EVC is caused by a misspelling in the *EVC* gene, which appears to play important roles in the normal shaping of many body parts. People affected with EVC generally have short stature (dwarfism; short arms and legs) and short ribs, which in turn causing narrowing of the chest that restricts air movement. About half of all babies with EVC syndrome are born with a life-threatening heart malformation that requires surgical repair. Other signs and symptoms include polydactyly (extra fingers or toes), missing or malformed nails, and dental abnormalities. The condition is inherited in an autosomal recessive pattern, meaning that a child diagnosed with EVC inherited two mutated copies of the *EVC* gene; one from each healthy carrier parent.

Information from NIH and GARD



L to R: **Dr. William G. Mackenzie & Dr. Stuart Mackenzie**

Collaborator Spotlight

William G. Mackenzie, MD and W.G. Stuart Mackenzie, MD

Department of Orthopaedic Surgery, Nemours Children's Hospital, Wilmington, Delaware

The father and son duo of Dr. William G. Mackenzie and Dr. Stuart Mackenzie regularly see patient families at the Clinic for Special Children as visiting specialists from the Department of Orthopaedic Surgery at Nemours Children's Hospital in Wilmington, Delaware. Dr. William Mackenzie is the Chairman of the Department of Orthopaedic Surgery and the holder of the Shands/MacEwen Endowed Chair at the Nemours Children's Hospital, Delaware. He is also a Professor of Orthopaedic Surgery at the Sidney Kimmel Medical College at Thomas Jefferson University. He earned his medical degree and completed his residency at the University of British Columbia and completed his internship at McGill University and completed his fellowship in pediatric orthopaedics at the Nemours Children's Hospital, Delaware.

Dr. William Mackenzie is also Chairman of the Medical Advisory Board for Little People of America and Medical Director of the Dwarf Athletic Association of America. His clinical interests include skeletal dysplasia, limb length discrepancy, and limb alignment. He is an active member in numerous societies and served on the Board of Directors for the Pediatric Orthopaedic Society of North America. His distinctions include being named "Best Doctors in America" and "Guide to America's Top Surgeons" since 2002. In his free time, he enjoys golfing, gardening, spending time with his grandchildren, and traveling.

His son, Dr. Stuart Mackenzie, is a Pediatric Orthopaedic Surgeon at Nemours Children's Hospital, Delaware. He serves as the Associate Fellowship Director at Nemours and is active in multiple national and international societies. A graduate from New York Medical College, he completed his orthopaedic residency at Columbia University, and a fellowship at Nemours. He is a member of the Nemours Skeletal Dysplasia team, providing comprehensive care to children with skeletal dysplasia, with a specific interest in spine deformity. He and his wife, Jil, have a three-year old daughter, Cora. They are very active and love to explore. He's an avid cyclist and can frequently be found on the roads of Lancaster County.

The Mackenzie's relationship with the Clinic dates back to when Dr. William Mackenzie was introduced to the Clinic by Dr. Freeman Miller, a longtime visiting specialist in Pediatric Orthopaedics from Nemours. Dr. Mackenzie started doing occasional clinics at the Clinic shortly thereafter. Today, Drs. William and Stuart Mackenzie visit the Clinic together every three months focusing on orthopaedic concerns for Clinic patients. "These are usually forms of skeletal dysplasia, most commonly Ellis-van Creveld and cartilage hair hypoplasia syndromes. We give guidance on non operative treatment frequently, and when a child does require surgery, we assist in their care at Nemours Children's Hospital, Delaware. We are fortunate to provide all follow up care at the Clinic," says Dr. Stuart Mackenzie.

When asked why they pursued a career in pediatric orthopaedics, Dr. Stuart Mackenzie explains, "I have always pursued a career in working with children, even prior to medical school, running educational programs for high school students. Even my wife is a teacher! I was very clearly influenced by my father, as well as time spent working in sub-Saharan Africa, witnessing pediatric conditions that were easily fixed through short surgeries. The opportunity to work with families, and improve the lives of children through orthopaedic surgery, makes my job the most rewarding profession in the world (to me)."

Dr. William Mackenzie replies, "It is fun working with children and other pediatric providers. I can have a lifetime impact on function for families with young children and follow them through school, jobs, marriage, etc."

For Dr. William Mackenzie, the proudest accomplishment of his career has been developing a good Orthopaedic Department and an amazing Skeletal Dysplasia team at Nemours Children's Hospital, Delaware. For Dr. Stuart Mackenzie, his proudest accomplishment to date has been "completing the 28 years of schooling to become a pediatric orthopaedic surgeon, but I still have plenty to achieve during my career. Being part of the world famous Nemours Skeletal Dysplasia team makes me very proud, and I look forward to helping continue to develop our program."

We are grateful to Drs. Mackenzie for providing comprehensive and compassionate care to families at the Clinic!

Our Staff

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Nurse & Cherished Lives Program Manager

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KaLynn Loeven
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Julia Martin
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L to R: **Carlyn Darby and Faye Brown**

Providing care from the heart

Meet nursing volunteers Carlyn Darby & Faye Brown

If you've visited the Clinic during one of our monthly immunization days, you've probably met Carlyn Darby and Faye Brown! Carlyn and Faye volunteer their time at the Clinic to help our nursing and clinical teams. "Following a prompt from a friend, I listened to a TEDx Talk by Dr. Kevin Strauss. I was excited to learn that the Clinic at which he worked was very nearby. I made the call into the Clinic to discuss the possibility of volunteering as a nurse, as I'm a retired pediatric nurse," explains Carlyn Darby of her initial interest in volunteering at the Clinic. Throughout her career, Carlyn worked in the hospital setting, as a missionary nurse in Appalachia, as a substitute school nurse, as an office nurse, and even as a clinical instructor.

"I heard about the Clinic for many years before reaching out to volunteer," explains Faye. "When I retired from a paycheck and a schedule five years ago, I wanted to continue working but in a volunteer role. I made a list of various opportunities in my area, which included the Clinic. After an interview with the staff, I knew there was a place for me to serve the community at the Clinic." Faye grew up in a mushroom farming community in Berks County and has been a registered nurse for 45 years. Her specialties during her career included critical care, trauma, and emergency nursing on adult and pediatric levels. She also taught at three area nursing schools as a pediatric clinical instructor. She has served as a missionary for 38 years and lived in Senegal, Africa for six and a half years.

Both Carlyn and Faye's favorite moments of volunteering at the Clinic involve meeting the families that visit. "I enjoy meeting the mothers and children who come to the community-based immunization clinic. I love watching the little ones grow and thrive in their families, it's a wonderful opportunity," says Carlyn. "My greatest joy is visiting with the parents and the children," explains Faye, "building relationships is so important, and we can learn so much from each other. Although I don't know Pennsylvania German to communicate with the little ones, they always return a smile... except right after they get a shot."

At the core of why Carlyn and Faye volunteer at the Clinic is a dedication to service towards others. Their work has helped the Clinic expand our vaccination program and has been essential to keep children and families well. We are so thankful to Carlyn and Faye for their steadfast and continued dedication to the Clinic's mission and to the families that rely on our services.

Scenes from Benefit Auctions

Enjoy the photos below from several of the 2022 benefit auctions!

Clinic for Special Children®

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2022 Extraordinary Give

Lancaster County's Largest Day of Online Giving!

Save the date for the 2022 ExtraGive on Friday, November 18th!
Last year over $110,000 was raised in 24 hours for the Clinic's mission.
Will you help us reach our goal of $100,000 this year?

Holidays at the Clinic

Our office observes the Thanksgiving, Christmas, and New Year holidays and will be closed or closing early on certain dates.
View the schedule on the cover of this newsletter for details.

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."