Clinic *for* Special Children

Newsletter

ISSUE #43

SPRING 2018

"God will provide direction and make a way when it seems like there is none. You are not alone."

– Joanna Weaver

A Story of Hope and Resilience: Against the Odds
The Weaver Family

When Ashton Weaver was one year old, he could stand by himself and play with toys like any other child his age. It seemed that Ashton was healthy, but gradually his legs began to hang limp, unable to support his weight. Through a diagnostic test in our laboratory, Ashton was diagnosed with Spinal Muscular Atrophy (SMA), a recessive genetic disorder that affects nerve cells housed in the spinal cord and brain, leading to a progressive loss of muscle function.

Over the subsequent six years, Ken and Joanna experienced the joyful births of three more boys: Lincoln, Brenton, and Winston. With the birth of each new child, came another positive SMA diagnosis. The probability of all four children inheriting SMA was 1/256 or less than half of one percent. Against the odds, the Weavers became parents to four boys living with the same rare, incurable, and previously untreatable genetic condition.

The Weavers turned to God for support. Joanna says, "God allowed them all to have SMA for a reason. For our own peace, we had to believe that and accept it, even when the future looked overwhelming!" The Weavers remained up-to-date with all of the new research and medical findings about SMA through a group called Cure SMA. Day to day, they leaned on the support offered by their church, their family, and friends.

Finally, in 2017, the Weaver's hope was restored. CSC's doctors became aware of a treatment option for SMA, a new drug that is injected directly into the fluid around the spinal cord, intended to increase motor function and extend life expectancy for those living with SMA. The CSC clinicians recommended that the Weaver boys participate in a clinical trial for this new treatment. To ensure that families could receive this treatment with no barriers to care, the CSC clinical team helped to enroll each child in a philanthropic program that would allow them to receive this medication, normally costing $125,000 per dose at three doses per year, for free for life.

All four boys were accepted into the program and have received at least four doses of the new drug. Today, you wouldn't know by the boys' infectious smiles and positive spirit that they were living with a crippling genetic disorder. Each boy's journey through this treatment is unique and the ultimate results of their treatments are unknown, but they are all full of new life and a new hope.

"This experience made us turn to God in a deeper way than anything else ever had, as we looked to Him for strength. We have learned to want His will above ours, even if it is not strong physical bodies for our children. If we could give a message to other families in similar situations, we encourage them to never give up on believing that treatment might be possible. Stay hopeful and do your own research to stay informed. The Clinic for Special Children is the most helpful place to go for the treatment of genetic disorders and we are very thankful for the caring and compassionate staff."

SPECIAL DATES

Auction Season is Here!
June - September
Find all auction dates, times, and locations on page 3

Patient Family Advisory Council Meeting
Tuesday, May 1
CSC, 7 p.m.-8 p.m.

CME Midwife Conference
Thursday, May 24
CSC, 8:30 a.m.-4:00 p.m. - Details inside

Properdin Family Day
Wednesday, June 27
CSC, 9 a.m.-5 p.m.

Transplant Family Day
Tuesday, July 17
CSC, 9 a.m.-5 p.m.

Nephrotic Syndrome Family Day
Tuesday, August 7
CSC, 9 a.m.-5 p.m.

CSC 1st Annual 5K RUN/WALK
Saturday, September 22
CSC, 8 a.m.-10 a.m

Patient Fall Family Fun Day
Saturday, October 13
CSC, 12 p.m.-4 p.m. - Details inside

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

Announcements from the Clinic

Staff News

Katie Williams, MD, PhD

Dr. Katie Williams will be leaving CSC in June 2018 to continue her pediatric practice in her home state of Wisconsin. We would like to express our gratitude to Dr. Williams for her four years of invaluable service to the Clinic for Special Children through her devotion to the patients and families she's served, her contributions to original scientific research and her passion for community education.

During her time with CSC, Dr. Williams collaborated with Dr. Devyani Chowdhury from Cardiology Care for Children to design and implement a globally recognized newborn wellness screening protocol. A speaker at Penn State University and PA Office of Rural Health's 2018 Community and Public Health Conference, she was able to present this project and research to an audience of medical practitioners, professors, government and state representatives, non-profit executives, and students. She has contributed original research to two articles published in peer reviewed journals with three others in progress. Her work and interviews have been featured locally in Lancaster Newspaper, Ephrata Review, and Advertiser, nationally in Genome Magazine, and internationally through Gulf News Magazine out of Dubai.

During her time with CSC, Dr. Williams passionately involved herself in the education and empowerment of midwives across Pennsylvania. She worked with CSC's Christine Hendrickson to organize three pulse-oximetry workshops for local midwives and they will host a CME accredited midwife conference next month at the Clinic.

Through her care for Amish and Mennonite children living with rare genetic disease in Lancaster, Dr. Williams created lasting relationships with many families in the community and developed a deep respect and devotion to those she has served. She hopes to continue her care of children from the Plain community in her home state of Wisconsin. We wish her all of the best in her next endeavours. Please find her handwritten note to all of you on page 8.

A New Research Operations Department at CSC

Patient focused research has always been an important component of CSC's mission. We strive to translate research and medical knowledge into practical benefit for the patient families we serve. In order to better support our programs, a Research Operations team has been established to manage and organize ongoing and future research.

Karlla Brigatti, MS, LCGC

Karlla has been appointed Research Operations Director and will lead the Research Operations team. Karlla will also join the leadership team of the Clinic.

Millie Young, RN

Millie has been appointed Research Nurse and will work with Karlla and the rest of the CSC team to achieve research goals.

Keturah Beiler, RN

Keturah has moved from a part-time nurse to a full-time nurse and will be providing clinical support to Dr. Strauss.

CSC Welcomes Development Director!

Teresa Rineer

In January, we welcomed a new member to the CSC team! As CSC's Development Director, Teresa is responsible for providing opportunities for support, stewarding philanthropic relationships, and serving on CSC's leadership team.

We hope these changes will further advance our mission of serving children who suffer from genetic and other complex disorders!

CSC 2018 Midwife Conference

Midwifery Pearls:
Caring for the Newborn

Thursday, May 24
8:30 a.m. - 4:00 p.m.
at Clinic for Special Children

This event is designed to provide midwives with the most up-to-date information, emphasizing recent developments in midwifery healthcare practice and to provide key solutions to the latest challenges faced by midwives. We also present a comprehensive review of disease screening and management for commonly encountered diseases in Amish and Mennonite communities. This course is intended for certified nurse midwives, lay nurse midwives, and others interested in the care of newborns. Register by May 18th through our website or call CSC for a paper application!

Patient Fall Family Fun Day!

Hosted by CSC's Patient Family Advisory Council

Saturday, October 13
12 p.m. - 4 p.m.
at Clinic for Special Children

Patients and their families are invited to join the CSC staff and board for a FREE day of fellowship and fun! Get to know the CSC staff, CSC board, supporting specialists, and their families! Enjoy activities at the Clinic between 12 p.m. and 4 p.m.: hayrides with antique tractors, children's activities, tours of the Clinic, delicious food, fresh cider, ice cream, and more! The event is hosted by the Patient Family Advisory Council (PFAC). The PFAC is a group of patients, family members, clinicians, and staff members that meet bimonthly with the purpose of incorporating a patient voice into the Clinic's programs and practices.

Save the Date for CSC's First 5K Run/Walk/Jog

Saturday, September 22
8 a.m. - 10 a.m.
at Clinic for Special Children

Please join us for our FIRST EVER 5K to benefit the Clinic for Special Children! Stay tuned to the 'Events' section of our website and our summer newsletter for details about our run as they become available.

2018 Auction Season is Near

Please join us this auction season for days of fellowship, family, and good food! Handmade quilts, handcrafted furniture, wooden crafts, sporting goods, handmade toys, and garden plants will be available to bidders at our auctions. A variety of food choices will be featured including chicken barbecue, freshly made pies, donuts, whoopie pies, ice cream and more! Proceeds directly support the mission of the Clinic for Special Children.

Making up approximately 60% of CSC's revenue, funds raised from our annual benefit auctions and charitable donations make it possible for the Clinic to continue to provide affordable, specialized care to children living with rare genetic disorders not just in Lancaster County, but world wide.

Please visit our website for the most current auction information.

Mark Your Calendar for our 2018 Benefit Auctions

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

Union County Auction - 6/2/18

Flower Sale and Rib Dinner - 6/1 - 5 p.m. - 8 p.m.
Buffalo Valley Produce Auction
22 Violet Road
Mifflinburg, PA 17844
Contact: Leon Hoover | 570-966-2414

Lancaster County Auction - 6/16/18

Leola Produce Auction
135 Brethren Church Road
Leola, PA 17540
Contact: Mark Martin | 717-733-3070

Shippensburg Auction - 6/23/18

Cumberland Valley Produce Auction
101 Springfield Road
Shippensburg, PA 17257
Contact: Elvin Oberholtzer | 717-532-9088

Blooming Grove Auction - 7/14/18

Blooming Grove Auction Inc.
1091 Free Road
Shiloh, OH 44878
Contact: Leon Newswanger | 419-896-3336

Blair County Auction - 9/8/18

Morrison's Cove Produce
174 Windy Acres Ln.
Roaring Spring, PA 16673
Contact: Paul Ray Fox | 814-224-5442

"Every day, we see families as they navigate a place they never expected to experience. Some are just starting their journey, some are many years into the journey. I try to imagine that family arriving in an unfamiliar place, and I wonder what would be helpful to them. Our goal is to provide comfort, guidance, and ease their journey, so that no matter where your journey takes you, you will not be alone."

– Katie Williams, MD, PhD

Flouride Varnishing

The importance of a healthy smile

By Donna Robinson, CRNP

You may have heard that your smile is the window to the world. Your smile is also a window to your overall health. Taking control of your child's health begins at an early age with a healthy diet, prevention of injury, preventative health check-ups, immunizations, and dental care. Taking care of your teeth and gums helps to prevent bacteria from entering other organs. Dental varnishing is one of the earliest things you can do to help your child develop strong, healthy teeth.

Dental caries (cavities) are the most common medical problem in children with many children developing caries by age six. Dental caries can lead to loss of teeth, dental infections, speech impairment, poor nutrition, decreased self-esteem, and school absences. Long term, poor oral health can lead to infective endocarditis, diabetes, obesity, heart disease, and adverse pregnancy outcomes. Children with special needs and intellectual disabilities are at higher risk for dental caries due to less than optimal conditions for good oral hygiene. Survey results published in the 2017 edition of *Journal of Clinical and Experimental Dentistry* suggest that approximately 88% of Amish children have untreated dental caries with almost all of them acquired by age 16. The good news is dental caries can be prevented.

The solution is a fluoride varnish. It is easy, inexpensive, safe, and proven to reduce the frequency and severity of dental caries in children. Dental varnishing should start as soon as teeth appear and should be applied two to four times per year through age five. The Clinic is offering dental varnishing for only $5 per child. It is quick and can be done during a routine office visit. For questions, please call the clinic and speak to your child's nurse.

GM3 Synthase Deficiency

A Family History Study

By Millie Young, RNC, Research Nurse

GM3 Synthase Deficiency is a disorder commonly observed in Amish communities which causes severe developmental delays, seizures, sleep disturbances, chronic irritability, hearing and vision problems. Although CSC has been working with partners to attempt to identify an effective medical intervention for many years, nothing has proven successful in providing significant treatment.

In the meantime, approximately 100 patients, living or deceased, with GM3 synthase deficiency have been identified in Pennsylvania, Ohio, Indiana, and Wisconsin. CSC is working with partners at the New Leaf Clinic in Ohio, Community Health Clinic in Indiana, and the Center for Special Children in Wisconsin to administer a survey to the families of affected children. The survey collects information about treatments used, severity of symptoms, and other important pieces of data. All of the information will then be summarized into a natural history study. Natural history studies are important because it gives researchers a detailed look at the natural course of a disease. Then if a medical intervention is attempted, the results can be compared to the data to see if the intervention is effective.

These studies allow us to learn from the people who know the disease best, parents and caregivers. We hope to be able to share the insights we learn with families as soon as data collection and analysis is complete. For more information, please contact Millie at the Clinic.

A New Service Available

SMA Carrier Testing

After months of development, the CSC laboratory is proud to announce a new carrier test for spinal muscular atrophy (SMA). In the Amish and Mennonite populations, SMA is caused by a homozygous deletion of the SMN1 gene. Affected children inherit one deleted copy from each parent (and thus have no functional SMN1). The new test uses competitive polymerase chain reaction (cPCR) to assess the number of copies of the SMN1 gene (versus a non-deleted gene, ALB) that an individual carries. SMA carriers harbor only one copy of the SMN1 gene. The new test will be offered for the same price as our other carrier tests ($50).

"Every deaf or hard of hearing child should have the opportunity to hear their parents say 'I love you.' "

-Dr. Thierry Morlet

Collaborator Spotlight: Thierry Morlet, PhD

Neuroscience, Auditory Physiology, and Psychoacoustics

DR. THIERRY MORLET, an accomplished neuroscientist and researcher, has been offering his services to the children served by CSC for the past six years. For over 12 years, Dr. Morlet has served as the head of the Auditory Physiology and Psychoacoustics Research Laboratory at Nemours/Alfred I DuPont Hospital for Children in Wilmington, Delaware. On a daily basis, he conducts basic and translational research on various types of hearing loss in children. Dr. Morlet enjoys the combination of his practice, as it enables him to acquire knowledge about diverse aspects of the human physiology, specifically including what can go wrong during human development. He also loves being able to combine his research studies with clinical activities. He says, "Interaction with patients is a constant reminder of the extraordinary application research can have. It helps reformulate research questions and, of course, is very rewarding when we can directly apply this new knowledge to improve the lives of children."

Dr. Morlet credits his original interest in pursuing a PhD in Neuroscience to his adolescent years in France. "When I was a young teenager, the different brain functions fascinated me. This is when I decided to become a 'brain' scientist, without knowing yet exactly what it meant. But when I left high school, I knew exactly what to do to get a PhD in Neuroscience." He became one of three postgraduate students to be accepted into a competitive PhD program in neuroscience at the University of Lyon in France. His courses of study exposed him to research in many areas of the brain, finding one laboratory director's description of paradoxical sleep state or REM particularly captivating.

Beyond his interest in neuroscience, Dr. Morlet found a particular passion for studying brain development in children. He says, "Studying how the brain develops to become this incredible 'machine' has always been one of my priorities. For example, the way most babies can learn language and how to speak by listening and observing their caregivers is fascinating. When I was a student, I had the opportunity to start studying the development of hearing in newborns. I jumped right in because I knew there was a lot to do for those who were not born with this essential sensory function."

More than six years ago, Dr. O'Reilly, an otolaryngologist working for Nemours, met with CSC's medical director, Dr. Kevin Strauss, and they started talking about a way to test hearing in the Plain population at the Clinic. Dr. O'Reilly and Dr. Morlet visited the Clinic and negotiated a collaborative agreement to provide audiology services on site. They started audiology testing for infants and children soon thereafter, using the existing sound booth at the Clinic and the equipment Dr. Morlet would bring with him. Thanks to this preliminary work, there is now an established monthly hearing clinic at CSC with auditory testing, hearing aid fittings, cochlear implants, and ENT services. "Our job is to help make sure even hearing impaired individuals can communicate with each other through hearing and spoken language whenever possible to thrive in the community."

Dr. Morlet says of his decision to collaborate with CSC, "I think that was a very easy decision for me as a scientist. There is so much to learn here and we knew that we could provide some help to the hearing impaired children served by the Clinic. It is a great opportunity to increase our understanding about why and how hearing and language function impairments coexist with some of the disorders seen by clinicians at CSC and it is rewarding to use our existing knowledge about hearing loss in infancy and the current ways we can manage it to impact the development of speech and language."

During his time working with CSC, Dr. Morlet has participated in several medical research studies, helped to provide clinical audiology services to patients living with rare genetic disease, and empowered families through educational days at the Clinic. He particularly enjoys participating in patient family days, "It is a great opportunity for us to share information with families about what we are doing, where we are in terms of research advances, what they can expect from us in the near future, etc. The most important aspect of those family days for me is that time spent dialoguing. I learn a lot from families and am hoping they are learning more about their child's disorder and how to best manage it for years to come. Time is always limited in a typical clinical setting. Here, during family days, time is not an issue."

When Dr. Morlet is not in the office, he enjoys traveling and photography. We appreciate the significant contributions that Dr. Morlet has made to CSC and the patients we serve.

Clinic *for* Special Children

Our Staff

Keturah Beiler, RN
Nurse

Karlla Brigatti, MS, LCGC
Director of Research Operations and Genetic Counselor

Kim Calderwood, MA
Communications Manager

Vincent Carson, MD
Pediatric Neurologist

Adam D. Heaps, MS
Executive Director

Christine Hendrickson, RNC
Nurse

Candace Kendig
Medical Receptionist

Lavina King
Community Liaison

Yalonda L. Kosek
Office Coordinator

KaLynn Loeven
Laboratory Technician

Erik G. Puffenberger, PhD
Laboratory Director

Teresa Rineer
Development Director

Donna L. Robinson, CRNP
Nurse Practitioner

Ashlin Rodrigues
Laboratory Technician

Kevin A. Strauss, MD
Medical Director

Katie B. Williams, MD, PhD
Pediatrician

Millie Young, RNC
Research Nurse

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Herman Bontrager
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Secretary, Chair-Development Committee

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Jacob Zook - *Vice Chairman*

Donor Spotlight

A Creative Approach to Advocacy

Esther Davies and John Thackrah beam with passion when they talk about the Clinic for Special Children. Through their creative approaches to education and advocacy for CSC over a combined 39 years, they have garnered invaluable support for the Clinic.

John has been an artist for as long as he can remember. His second painting from 1951 is centered over his couch pictured above. Over the past 40 years, John has used nature, wildlife, buildings, and landmarks from across several states and countries as inspiration for his artwork.

Esther's support of CSC's mission stems from her very close ties to the Amish community for over 35 years. John and Esther met almost nine years ago when John and his wife, Doris, moved into Willow Valley Communities. They formed a friendship immediately. Esther invited John to tour CSC and when John saw the Clinic firsthand, he knew he had to do something to help.

John and Esther arranged a creative awareness campaign for CSC. John converted his pen and ink artwork depicting native and iconic landmarks from around Lancaster County into note cards and matted prints. Proceeds raised from the sales of John's cards and prints benefit CSC. Esther and John work with volunteers from Willow Valley to box the cards and pictures for sale.

Through card sales alone, John, Esther, and volunteers from Willow Valley have raised over $3,000 for CSC. Additionally, their public support through advocacy and education of others in Lancaster County about the Clinic's work and mission has lead to critical support for CSC. The Clinic for Special Children welcomes and is grateful for creative ideas that support the Clinic and increase awareness of CSC's work.

We are honored to present this story about the incredible impact one beautiful woman, Esther Davies, made on the Clinic for Special Children and the Plain communities through a lifetime of passion and advocacy. We are now sharing her story with a heavy heart as Esther passed in March, weeks after this photograph was taken. We invite you to help us celebrate her legacy by sharing her story of compassion. On behalf of the board and staff at CSC, and the countless children who have benefitted from her kindness, we wish to express our sincere gratitude and condolences to Esther's family and friends.

Development and Outreach

"I'm constantly inspired by the generosity of CSC's giving community and truly grateful for the opportunity I've had to build meaningful relationships with many of the special children we serve and their families. One such opportunity lead Dr. Strauss and I to Arizona to support the Crain Family Foundation's 'Swing FORE the Kids' Topgolf event to benefit CSC (pictured right). I look forward to providing giving opportunities for all of those who believe in the mission of CSC and are moved to contribute."

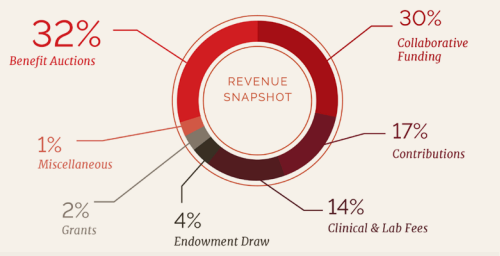
-Teresa Rineer
CSC Development Director

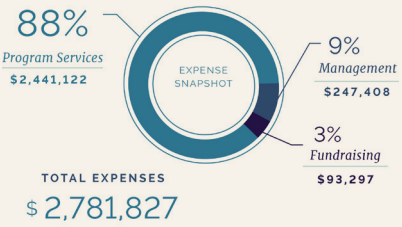
The Clinic is funded from four major sources of revenue — annual auctions, collaborative relationships, clinical and laboratory fees, and donations from caring people just like you.

We Need Your Support

Ways you can support the Clinic

1. **Support our auctions and special events:**
   - Attend our special events and invite others
   - Donate items to our auction
   - Be a sponsor or volunteer for a special event
2. **Make a monetary donation**
   - Show your support through the year-end appeal
   - Give anonymously through Anabaptist Foundation
3. **Participate in our planned giving program**
   - Include CSC in your estate plan, will or trust
   - Designate CSC as a beneficiary of your IRA
   - Donate stock or farm commodities





We work hard to keep clinic fees to a minimum to ensure the families we serve can afford the consistent care they need. Amish and Mennonite communities work together to raise funds by hosting auctions that take place across Pennsylvania and Ohio. These events are a time of fellowship for the communities we serve and remind everyone of the importance of our mission. Private philanthropic donations allow us to conduct research, provide community education, and coordinate patient care. Your support enables CSC to continue to provide patients with timely, affordable, and effective care!

International Outreach

CSC Mission to Mexico

Three thousand miles is a long way to travel to make house calls. But that's what Medical Director Dr. Kevin Strauss, Genetic Counselor Karlla Brigatti, and Board Chairman Herman Bontrager did last September when asked to provide medical attention to members of Old Colony Mennonites in Mexico's Yucatan region.

The request came from the Anabaptist Foundation, which helps support the work of Mexico Mennonite Aid (MMA). Richie Lauer, Foundation Officer for the Anabaptist Foundation, contacted the Clinic for Special Children. He explained, "We reached out to the Clinic for Special Children because of their work in treating rare genetic disorders among Amish and Mennonite children, and their understanding of Plain community culture."

During their visit, Dr. Strauss and Karlla recommended treatment or follow-up analysis for 32 patients, both children and adults, for a wide range of conditions and identified ways to help others. They also provided guidance to MMA for the new maternity clinic being built for the colony.

Herman explained, "Our goal was to respond to Mexico Mennonite Aid's need to provide health assessments and determine what, if any, inherited genetic disorders are a factor," he said. "We also offered to share our knowledge and experience in supporting them in developing their own clinic."

A Letter to Patients and Families

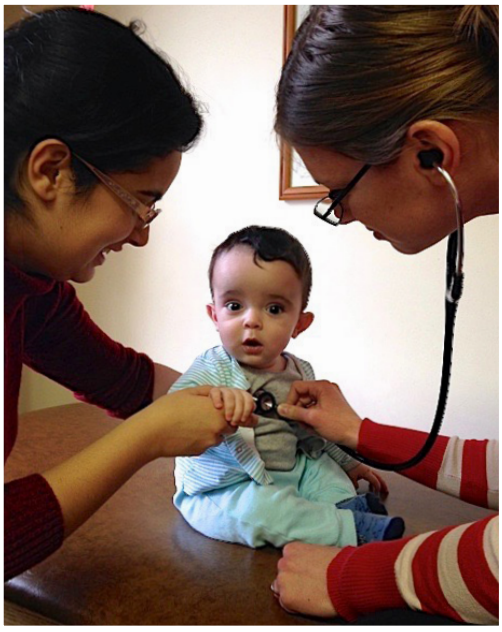
From Dr. Katie Williams

Dear patients and families,

When I arrived at the Clinic for Special Children four years ago, I did not know what to expect. I was new to medical practice, but eager to learn and contribute to the Clinic's mission. What I found at the Clinic exceeded all of my hopes; dedicated staff, loving families, and countless children that have become dear to me.

Work at the Clinic has enriched my life in many ways, but requires that I live a great distance from my extended family in Wisconsin. My husband and I share fond memories of childhood with our grandparents, aunts, uncles, and cousins, and want our children to have the same opportunity. After much thought and consideration, we have decided to return to Wisconsin to be closer to our families.

The Clinic has nurtured a deep, personal commitment to serving the Plain community. Moving forward, I hope to care for Amish and Mennonite children with genetic disorders and special medical needs in my home state. I will begin with a 2-year training program in medical genetics at the University of Wisconsin. This will give me the opportunity to do outreach work with the Plain communities in Wisconsin and share the knowledge that I have gained here in Pennsylvania.



I will be leaving the Clinic in June 2018. During the upcoming months, I plan to work closely with the Clinic staff to ensure a smooth transition of care for patients. As we work through this process, please let me know if there is anything I can do to help.

This change is bittersweet. Although I look forward to sharing more time with my extended family, I will greatly miss the staff and families at the Clinic. Each relationship, no matter how long or short, has shaped me as a doctor and as a person. Thank you for allowing me to be part of your lives in such a special way. Caring for your child has been a true pleasure and a gift I will cherish.

Sincerely,
Katie Williams

Newborn wellness screen helps save local life:

Pulse-Oximetry

Preston Groff Photography

Through partnerships between Clinic for Special Children (CSC), Cardiology Care for Children (CCC) in Lancaster, and midwives throughout the region, babies born at home are now receiving vital wellness screenings and, if a problem is detected, getting immediate medical care before they become critically ill.

The test, called pulse oximetry, measures the newborn's oxygen levels to detect heart defect, lung disease, and infections. Mallory Sensenig of Denver realized first-hand the importance of the screening when, within hours of delivering her daughter, Josephine, her nurse midwife, Danielle Malik, found the newborn's oxygen levels below normal.

The little girl was immediately seen by pediatric cardiologist Dr. Devyani Chowdhury of CCC. Dr. Chowdhury did not detect any heart problems and recommended the baby to Heart of Lancaster. Baby Josephine was admitted to the Neonatal Intensive Care Unit where she was treated for fluid in her lungs. After two days, her parents were able to take her home to her four brothers and sisters. On Jan. 25, Josephine celebrated her one-year birthday and is a normal, healthy toddler.

The at-home wellness screening program was developed in 2015 by CSC's Dr. Katie Williams and Dr. Chowdhury after PA state law mandated, in 2014, that pulse oximetry screenings be performed at 24 to 48 hours of age as part of every newborn's physical examination regardless of their place of birth.

Dr. Chowdhury's protocol, based on models used in Europe and the Netherlands where there are many home births, requires screenings be performed before the midwife leaves the mother and the baby, typically when the baby is three to five hours old, and a second screening per the state requirement from 24 to 48 hours after birth. "Dr Williams says, "Dr. Chowdhury developed the screening protocol and worked with the Clinic to find initial funding to purchase the pulse oximeters. We held our first workshop to train midwives on using the equipment in 2015."

Since then, nearly 2,000 newborns have received wellness screenings by midwives. As a result of the in-home screenings, seven newborns have been identified with heart or lung problems and received care before potentially becoming critically ill.

RECENTLY PUBLISHED PAPERS

Yang Tan T, Gonzaga-Jauregui C, Bhoj EJ, Strauss KA, Brigatti KW, Puffenberger E, Li D, Xie L, Das N, Skubas I, Deckelbaum RA, Hughes V, Brydges S, Hatsell S, Siao C, Dominguez MG, Economides A, Overton JD, Mayne V, Simm PJ, Jones BO, Eggers S, Le Guyader G, Pelluard F, Haack TB, Sturm M, Riess A, Waldmueller S, Hofbeck M, Steindl K, Joset P, Rauch A, Hakonarson H, Baker NL, Farlie PG. **Monoallelic BMP2 Variants Predicted to Result in Haploinsufficincy Cause Craniofacial, Skeletal, and Cardiac Features Overlapping Those of 20p12 Deletions.** Am J Hum Genet. 2017 Dec. 101(6):985-994.doi:http://dx.doi.org/10.1016/j.ajhg.2017.10.006.

Chowdhury D, Williams KB, Chidekel A, Pizarro C, Preedy C, Young M, Hendrickson C, Robinson DL, Kreiger P, Puffenberger EG, Strauss KA. **Management of Congenital Heart Disease Associated with Ellis-van Creveld Short-rib Thoracic Dysplasia.** The J of Pediatr. 2017 Dec. doi: 10.1016/j/jpeds.2017.08.073.

Strauss, KA, Gonzaga-Jauregui C, Brigatti KW, Williams KB, King AK, Van Hout C, Robinson DL, Young M, Praveen K, Heaps AD, Kuebler M, Baras A, Reid JG, Overton JD, Dewey FE, Jinks RN, Finnegan I, Mellis SJ, Shuldiner AR, Puffenberger EG. **Genomic diagnostics within a medically underserved population: efficacy and implications.** Genet Med. 2017 July 20. doi: 10.1038/gim.2017.76.

InokuchiJI, GoS, Yoshikawa M, Strauss KA. **Gangliosides and Hearing.** Biochim Biophys Acta. 2017 May 30. pii: S0304-4165(17)30177-0. doi: 10.1016/j.bbagen.2017.05.025.

Aubert G, Strauss KA, Lansdorp PM, Rider NL. **Defects in lymphocyte telomere homeostasis contribute to cellular immune phenotype in patients with cartilage-hair hypoplasia.** J Allergy Clin Immunol. 2017 Jan 23. pii: S0091-6749(17)30055-6. doi: 10.1016/j.jaci.2016.11.051

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clinicforspecialchildren.org

2018 Benefit Auctions for Clinic for Special Children

June 1 & 2 - Union County Auction
June 16 - Lancaster County Auction
June 23 - Shippensburg Auction
July 14 - Blooming Grove Auction
September 8 - Blair County Auction

Dates, times, and locations inside!

"Every day, there is a growing need for the Clinic, but also for growing support. This safe haven extends beyond the Clinic staff, beyond the Clinic walls, and into the surrounding community. All this to ensure that your children, and your children's children, will always have a place to go."

- Katie Williams, MD, PhD