



Clinic for
Special Children

Newsletter

VOLUME 2 NUMBER 1

FALL 2016



Patient Focus:

A Medical Home

Alton and Sharla Zimmerman are parents to four beautiful children: Jennica, Kylen, Benjamin, and Lucia (pictured above L-R).

When Jennica was about two years old, she started to display symptoms that something may be wrong: seizures, speech difficulty, and unexplained behaviors. Sharla sought help from many resources in the community and spent many hours a day following grueling regimens and treatment plans that had short-term benefits to Jennica's health. When Kylen was an infant, he began showing symptoms similar to his sister which lead Sharla to seek care from the Clinic for Special Children. After meeting Dr. Strauss, Sharla reflects that she "felt like I could take some of it off my shoulders. He really respected a mother's intuition."

At the Clinic, the Zimmermans took part in a family study which mapped their genealogical history and analyzed their DNA. The children were both diagnosed with Cortical Dysplasia - Focal Epilepsy (CDFE or CASPR2), a rare, recessive, disorder which is linked to epilepsy, language regression, and autism.

Following their diagnosis, Kylen and Jennica were put on a new, less intensive medical and dietary regimen. Since they began treatment at the Clinic, both children's seizures are under control. They are vibrant and healthy, able to enjoy school and be active at home.

Sharla wonders, "If the Lord also gave Kylen CDFE so that Jennica could come to the Clinic." She feels that the Clinic has become her family's medical home. Now, after her children's diagnosis, Sharla still tries to learn all she can about the rare genetic disease so that she might help other struggling families. The Zimmermans are regular guests at the Clinic's educational family days and recently attended an event specifically for families impacted by CDFE. Sharla enjoys these events and meeting people who have gone through similar situations. Most of all, she enjoys the feeling of not being alone.

"I needed somebody to walk with me and I needed a safe place to go. If my experience can help someone else, I believe that is God's plan for my life."



SPECIAL DATES

Family Bereavement Day
October 12

CSC, 9am-4pm

Midwife Pulse-Ox Workshop
October 13

CSC, 1pm-4pm

5k, 10k, and Kids's Fun Run
October 22

New Holland, 8am Registration

Willow Valley Craft Show
November 18

Willow Valley Communities, 10am-4pm

Extraordinary Give
November 18

Lancaster, All Day Event

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



Announcements From the Clinic



It's Flu Shot Season

Flu shot season will be upon us soon. Please call the office at 717-687-9407 to schedule

your flu shot. Anyone over six months of age is encouraged to get a flu shot. We offer shots to our patients and their immediate family. The cost is \$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.

Get Involved with our New Patient Family Advisory Council

CSC has long collaborated with our patients and the Plain communities in order to provide comprehensive local medical care, integrate science and clinical medicine, and share knowledge to improve the health of children who suffer from genetic disorders. CSC's Board recently approved the creation of a Patient Family Advisory Council (PFAC), which is intended to partner patients, families, and caregivers with members of the CSC clinical team to collaborate on how to improve our patient and family experience. The council will meet bi-monthly to share their points of view, perspective, and experience for the purpose of ensuring CSC continues to provide the highest standards of comprehensive, compassionate, and culturally sensitive health care. If you have a patient history with CSC, either by being a patient yourself or a family member of a patient, and you are interested in learning more about this group, please contact Kim Calderwood at 717-687-9407 or kcalderwood@clinicforspecialchildren.org.



Join us for Boy Scout Troop 48's 5k, 10k, and Kid's Fun Run for CSC!

Please join Boy Scout Troop 48 and CSC on

Saturday, October 22, 2016 for a 5k, 10k, and Kids Fun Run at the Welsh Mountain Park. The run is through country roads with a great autumn view of the valley. Entrants are requested to bring 2 non-perishable food items along with the \$15 fee for 5k, \$20 fee for 10k, and \$5 fee for the kid's fun run. Proceeds benefit the Clinic for Special Children and New Holland Boy Scout Troop 48! This is an event for all ages and any running level! The day's events are planned as follows:

Registration: 8am-10am
Kid's Fun Run: 9:30am
5k and 10k Run/Walk: 10am

Applications can be found at www.clinicforspecialchildren.org

To request a copy of the application to be mailed to your home, please call our office at 717-687-9407.

Mail written applications to:
Troop 48
20 South Roberts Avenue
New Holland, PA 17557

OR

Online registration is available at Active.com.



3rd Annual Whoopie Pie Toss for Lancaster's Extraordinary Give!

The countdown to the 2016 Extraor-

dinary Give is on! On **November 18, 2016** donations will be accepted from 12 midnight - 11:59 PM as part of Lancaster County's **largest** day of online giving. Every dollar donated at ExtraGive.org on November 18 will be stretched by a pool of at least \$300,000 from the Lancaster County Community Foundation, Rodgers & Associates, and other corporate sponsors AND there are at least \$50,000 in EXTRA prizes you can win for the causes you care about.

Donating is simple. On **November 18, 2016** visit ExtraGive.org and select 'Clinic for Special Children' as your **non-profit organization of choice**. If you would prefer to donate over the **phone**, please call the Clinic on November 18th between the hours of 9am and 5pm and we would be happy to receive your gift over the phone.

In 2015, nearly 60% of our revenue came from charitable contributions and benefit events.

Visit us on November 18th in Lancaster City at the Citizen's Bank Quadrant across from Lancaster Central Market for our 2nd Annual Whoopie Pie Toss in celebration of the ExtraOrdinary Give!

For additional information, please contact Christine Stickler at 717-687-9407 or cstickler@clinicforspecialchildren.org.

Benefit Auction Recap

We would like to extend a heartfelt thank you to everybody that helped to make our benefit auction season an overwhelming success this year. Thank you to our auction committee members, volunteers, donors, and all of our supporters who came out to enjoy the auctions and join in a day of fellowship, family, and good food!



Mark Your Calendar for our 2017 Benefit Auctions

Union County Auction - 6/3

Buffalo Valley Produce Auction
22 Violet Road
Mifflinburg, PA 17844

Lancaster County Auction - 6/17

Leola Produce Auction
135 Brethren Church Road
Leola, PA 17540

Shippensburg Auction - 6/24

Shippensburg Auction Center
1120 Ritner Highway
Shippensburg, PA 17257

Blooming Grove Auction - 7/8

Blooming Grove Auction Inc.
1091 Free Road
Shiloh, OH 44878

Blair County Auction - 9/9

Morrison's Cove Produce
174 Windy Acres Ln.
Roaring Spring, PA 16673



"It is not enough to merely care for your patients, you must also care about them. When you start caring about them, they demand your time and effort; they begin to shape your future. The hours of worry are not a burden—they humble us, strengthen us, and make us mindful of the needs around us. They teach us compassion—which literally means 'to suffer with.' Dr. Kevin Strauss, CSC Medical Director



CSC's First Pediatric Neurologist: Vincent Carson, MD

As the number of patients under the Clinic's care grows, it is important to ensure that our clinical staff grows in parallel. We are proud to announce that Dr. Vincent Carson joined CSC's team in July 2016. Dr. Carson committed to becoming a member of CSC's staff in the summer of 2015 during his last year of residency. We remain dedicated to providing the highest quality of clinical care for our patients. By attracting top medical professionals to CSC, we will continue to fulfill our mission for many years to come.

Dr. Carson is a pediatrician and child neurologist. As a neurologist he specializes in disorders of the brain, spinal cord, nerves, and muscles.

"I am honored to join the staff here at CSC and serve children suffering from rare genetic disorders."

Personally, he has a special interest in the genetics of brain disease. He earned a bachelor's degree in Biochemistry and Molecular Biology from Penn State University in 2004 and a medical degree from Drexel University in 2011. Dr. Carson recently

completed his pediatric and child neurology training at the Children's Hospital of Pittsburgh. He has affiliations with the American Academy of Neurology and the Child Neurology Society.

Dr. Carson, his wife Jen, and their four children were excited to move back to their native Lancaster County, which brings them close to their extended family again. When he isn't seeing patients or with his family, you may find Dr. Carson playing ice hockey on a local community team!

Cutting-Edge Research: CDFE Project

Comprehensive clinical care and an on-site biochemical and molecular genetic testing allow the Clinic for Special Children to conduct patient-focused translational research. In the more than 20 years since its founding, CSC staff have published more than 100 peer-reviewed research papers, fueled by close internal collaboration between clinical and laboratory staff and effective cooperation with academic, scientific, and clinical partners. CSC is dedicated to the idea that genomics can be integrated into local medical care to reduce childhood death, disability, and chronic illness. The ultimate goal of all research at CSC is to apply advanced molecular genetic techniques to the care of individual patients and thereby improve regional public health.

Cortical Dysplasia-Focal Epilepsy (CDFE) Syndrome is an autosomal recessive disorder caused by variants in the *CNTNAP2* gene. The variant is commonly present in the Old Order Amish population. The *CNTNAP2* gene encodes Contactin-Associated protein-like 2 (CASPR2), which is important for neurodevelopment. Children with CDFE syndrome develop focal seizures in early childhood that are often resistant to medical management. The majority of these children also have intellectual disability and autistic features with behavioral issues that are challenging to treat.

In summer 2016 we began a study with the purpose of characterizing the full spectrum of symptoms of CDFE syndrome in order to appreciate the range of disease severity as well as the unique challenges that these families face on a daily basis. In order to accomplish this task, we are performing a retrospective review of patients with CDFE syndrome. Assessments include growth parameters, seizure characteristics, developmental milestones, medication use, as well as of the results of brain MRIs and EEGs. In addition, we are interviewing the parents of children with CDFE syndrome using validated surveys to thoroughly assess different issues, such as adaptive behavior and the psychological well-being of caregivers.

This study will help us to fully appreciate the natural range of variability of CDFE syndrome, which will allow us to adequately assess the efficacy of treatments in the future.

by: Vincent Carson, MD

OUTREACH AND RESEARCH

Williams, Katie. **A Dental Exam Reveals a Boy's Rare Disorder**. Interview by Sarvy Geranpayeh. Weekend Review. Gulf News, 18 Aug. 2016. Web.

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Herrick JR, Paik T, Strauss KA, Schoolcraft WB, Krisher RL. **Building a Better Mouse Embryo Assay: Effects of Mouse Strain and In Vitro Maturation on Sensitivity to Contaminants of the Culture Environment** AReprod Genet. 2016 Feb;33(2):237-45. doi: 10.1007/s10815-015-0623-y.

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Motzek A, Knezevic J, Switzeny OJ, Cooper A, Baric I, Beluzic R, Strauss KA, Puffenberger EG, Mudd SH, Vugrek O, Zechner U. **Abnormal Hypermethylation at Imprinting Control Regions in Patients with S-Adenosylhomocysteine Hydrolase (AHCY) Deficiency**. PLoS One. 2016 Mar 14;11(3):e0151261. doi: 10.1371/journal.pone.0151261. eCollection 2016.



An International Influence in Cardiology Care Dr. Sergi Cesar

The Clinic for Special Children would like to thank Dr. Sergi Cesar for his fantastic and selfless work at our Clinic. While researching new forms of heart diseases in the Amish and Mennonite communities, he generously volunteered his time to provide much needed care to the patients of the Clinic. His work, has saved Plain community families hundreds of dollars in cardiology (heart) screenings while performing research that could save lives globally.

A resident of Tarragona, Spain, Dr. Cesar came to the United States to study echocardiography with Dr. Devyani Chowdhury, a pediatric cardiologist practicing in Lancaster who donates her time and talents to see patients at CSC. Echocardiographs (commonly called an echo) are 2-D pictures of the heart made with ultrasound waves used to discover heart diseases. Dr. Cesar says that he fell in love with the investigative nature of echoes during his studies at Barcelona Saint Joan de Deu. He says that analyzing echoes are, "like solving a mystery, like Sherlock Holmes."

In addition to practicing echography, Dr. Cesar came to the states to study a cardiomyopathy caused by variants in desmoplakin (DSP) in the Amish communities of Dover, DE. These variants can cause cardiomyopathy (heart muscle disease), the symptoms of which are shortness of breath, tiredness, heart

failure, arrhythmia, or in the worst case, sudden death.

Working with the Amish community in Dover, Dr. Cesar was able to perform echoes on 55 patients in 10 days using equipment partially paid for by the Clinic. These echoes will form a study sample that will be the foundation of his research. Although it will take a while to analyze every detail of the echoes, he is optimistic that his findings will be published. His supervisor, Dr. Chowdhury had nothing but praise for his work ethic and dedication. "He has pushed our research," she declared. "He does high-quality work and is a great student." All in all, Dr. Cesar has clocked in approximately 400 hours of echo analysis since he came to the states. That totals about 170 echoes in all.

This was the first time Dr. Cesar visited the United States and worked with an organization like CSC. Ultimately, he spent 2½ months in the country. He believes that "The Clinic is a model for providing high-quality care." His contribution of time and effort helps to make that statement a reality.

Dr. Sergi Cesar is returning to Spain, but he will continue to collaborate with our team in Lancaster about his research in cardiomyopathy. From all of us here at the Clinic, thank you.

by: Liel Pollock

"Dr. Weiner has performed vital knee surgery for three of our children with EVC. The surgeries changed their lives and we are very thankful. I don't know where our family would be without him."

- AS, Father of 3 Patients



Dennis Weiner, MD

10 Years of Service to CSC

We would love to extend a deep thank you to Dr. Dennis S. Weiner, MD as we celebrate his 10 years of service to the patients of the Clinic for Special Children. Dr. Weiner is an orthopedic surgeon based out of Akron Children's Hospital in Ohio who specializes in knee and hip repairs for children with dwarfism. The two disorders we see most often at CSC that cause dwarfism are cartilage-hair hypoplasia (CHH) and Ellis-van Creveld syndrome (EVC).

Ten years ago, Dr. Weiner had the chance to evaluate a Lancaster County family in Akron. After considerable time spent with that family, Dr. Weiner performed vital knee surgery to three of their children with dwarfism. In order to follow up with the patients, Dr. Weiner traveled to Lancaster County and saw the family in a hotel room at the Best Western in Intercourse, PA. It was then that Dr. Weiner learned about the Clinic for Special Children and reached out to the organization to begin collaborating to provide services to children with rare genetic diseases.

To volunteer at the Clinic, Dr. Weiner and his nurse, Shannon, make a **five-and-a-half-hour drive** from Akron Ohio, where he practices, to Strasburg Pennsylvania twice per year. The long drive is worth it for both Dr. Weiner and his nurse, "It has been a personal pleasure for me and for Shannon...to come to the clinic in Strasburg and provide services for patients with these unique genetic disorders." Dr. Weiner's services have been life-changing for the patients of CSC, allowing them to receive the pain-relieving surgical services and consultations or providing transport to Akron's state-of-the-art Skeletal Dysplasia Center. Dr. Weiner has generously **never charged** our patients for his services at CSC.

Dr. Weiner has been able to compile the information he gathered over the 10 year period he has volunteered at CSC in order to publish his findings in the Journal of Children's Orthopedics. Specifically he has contributed to detailing operative approaches to 'severe knock-knees' deformities present in patients with EVC and CHH.

Dr. Weiner would like to express his enormous respect and admiration for the CSC clinical team for the "incredible service they have contributed to the care of so many Plain people through the clinic in Strasburg. It is an honor to be a consultant part of that group. I look forward to many visits in the future."

We wish to express our thanks to Dr. Weiner for his phenomenal work and partnership!



Our Staff

Karlla Brigatti, MS
Genetic Counselor

Kim Calderwood, MA
Communications Manager

Vincent Carson, MD
Pediatric Neurologist

Adam D. Heaps, MS
Administrative Director

Christine Hendrickson, RNC
Nurse

Yalonda L. Kosek
Medical Receptionist

Mindy Kuebler, MS
Laboratory Technician

Stephanie Kulp, RN
Nurse

Erik G. Puffenberger, PhD
Laboratory Director

Donna L. Robinson, CRNP
Nurse Practitioner

Kevin A. Strauss, MD
Medical Director

Christine Stickler, JD
Development Director & General Counsel

Carolyn K. Williams
Laboratory Technician

Katie B. Williams, MD, PhD
Pediatrician

Millie Young, RNC
Nurse

Our Board of Directors

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Enos Hoover

Leonard Hurst

Mark Martin
Treasurer

Caroline Morton, EdM
Secretary

Stephen D. Ratcliffe, MD, MSPH

Jacob Zook

Patient Artwork

We are now featuring
YOUR work! If you have
poetry, a letter to the Clinic,
or artwork that we may
feature in our next newsletter,
please send it to:

535 Bunker Hill Road
Strasburg, PA 17579



*"Have a hearty
handshake for me!"*

- John Mark & Ella Mae

I have a favorite Doctor
He lives in the country town
the Clinic is among the Amish
He likes to bike up hill and down

I love to visit him alot
though they like to pick on me
they are all so loving and kind
All of them I love to see

I do not know what I would do
If I would not have all of you
You are all so fantastic friends
I thank God for all of you!



*"Thank you. You
make me smile."*

- Love, Kierra



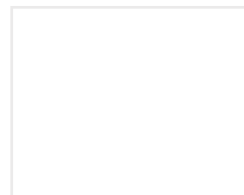
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Clinic for Special Children 2016 Staff

Front (L-R): Kim Calderwood, Christine Hendrickson, Vincent Carson, Karlla Brigatti, Donna Robinson, Carolyn Williams
Back (L-R): Adam Heaps, Yalonda Kosek, Millie Young, Kevin Strauss, Katie Williams, Christine Stickler, Stephanie Kulp, Erik Puffenberger
Not Pictured: Mindy Kuebler