



Clinic for
Special Children

Newsletter

ISSUE #48

FALL 2019



One in a million The Rosebrook Family

Brittany Rosebrook noticed something wasn't quite right during her pregnancy when she didn't feel her son Tanner kicking or moving actively. After Tanner was born at the hospital, he was sent for a car seat test and was promised to be returned within an hour. This test places the baby in a properly reclined car seat while nurses monitor their breathing and respiration to make sure they can safely travel if they have weak airways. After more than an hour passed, Brittany learned that Tanner started having seizures during the test and was transferred to a different hospital's NICU.

Tanner's initial hospital stay lasted two weeks and was riddled with countless tests and labs that all came up inconclusive. When the family finally arrived home, Tanner developed severe dystonia of all extremities that required emergency medication. The episodes occurred multiple times a day, lasting up to three hours each, and required rescue medications to end them.

At five months old, Tanner went to a local children's hospital for evaluation of severe dystonia and received the genetic diagnosis of Alternating Hemiplegia of Childhood. This is an extremely rare disorder where half of the body can be temporarily paralyzed. In addition, involuntary muscle movements can

also happen. The Rosebrook family finally had a diagnosis, but Tanner's journey was still a mystery. Alternating Hemiplegia of Childhood affects about one in a million people, and Tanner's specific genetic variant, in the gene *ATP1A3*, had never been reported before. The variant is *de novo*, meaning it was a random event and not inherited from either parent.

Brittany explains that after Tanner's diagnosis, they went to a neurologist and other specialists before finally visiting Dr. Devyani Chowdhury at Cardiology Care for Children in Lancaster, PA. Once Dr. Chowdhury saw Tanner, she told the family that they needed to go to the Clinic for Special Children to see Dr. Vincent Carson.

Brittany distinctly remembers the first time Tanner visited the Clinic, Dr. Carson exclaimed, "He's so handsome!" Dr. Carson's goal after this first visit was to provide Tanner the best quality of life possible. He worked with the family to reduce the amount of medications Tanner was on and implement an optimized nutrition plan. Brittany explains, "Dr. Carson has provided us a life and we're not in constant survival mode anymore. There's this fearless neurologist that sees Tanner's potential and wants to get there. He treats Tanner like a human and not a subject."



SPECIAL DATES

The Great Train Robbery Saturday, October 12

Strasburg Rail Road

www.StrasburgRailRoad.com

Portion of ticket sales benefits CSC

2019 Extraordinary Give Friday, November 22

Online Fundraiser

All Day - Details Inside

Thanksgiving - Office Closed November 28 & 29

All Day

The Night Before Christmas Train December 5-23

Strasburg Rail Road

www.StrasburgRailRoad.com

Portion of ticket sales benefits CSC

Christmas Eve - Office Closed Tuesday, December 24

1 p.m. - 5 p.m.

Christmas Holiday - Office Closed December 25 & 26

All Day

New Years Eve- Office Closed Tuesday, December 31

1 p.m. - 5 p.m.

New Years Day- Office Closed Wednesday, January 1

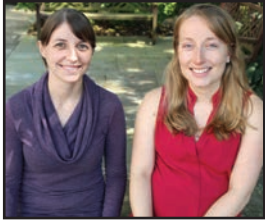
All Day

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

Staff News

KaLynn Loeven & Ashlin Rodrigues

Join us in congratulating Ashlin Rodrigues and KaLynn Loeven who were promoted to Laboratory Scientists! Ashlin and KaLynn both work tirelessly in our laboratory every day. We're thankful for their contributions to the Clinic for Special Children (CSC) team. Congratulations, Ashlin and KaLynn!



Dr. Stephen Ratcliffe

Dr. Ratcliffe joined CSC's board in 2014 while he was the Program Director of the Penn Medicine Lancaster General Health Family Practice Residency Program, a position he held between 2002-2019.

He is now shifting from the board to CSC's staff, serving as a Senior Consulting Physician. This part-time role will focus on the development of adult services at CSC, helping with educational efforts and scholarly research. We're excited to welcome Dr. Ratcliffe to our staff!



Emily Seitz

Emily Seitz is starting as the full-time Development Director in October. Emily joined the CSC team in August 2018 as a part-time Scientific Grant Writer. She is currently a PhD candidate at Penn State.



Emily has been instrumental in formalizing our grant writing program, and we're excited to have her in the expanded role of overseeing all fundraising efforts. Congratulations, Emily!

Board Updates

Peter B. Crino, MD, PhD

Dr. Crino is joining the Clinic's Board of Directors in October. He is an internationally recognized physician-scientist specializing in developmental brain disorders.

Dr. Crino has conducted extensive research on STRADA (Pretzel Syndrome) and is currently working with Dr. Vincent Carson on researching seizure-causing mutations (NPRL3 and DEPDC5). He currently serves as Chair of the Department of Neurology at the University of Maryland School of Medicine. Welcome, Dr. Crino!



Richard A. Fluck, PhD

Richard A. Fluck, PhD is retiring from the Clinic's Board of Directors in October. Dr. Fluck joined the CSC Board in the Spring of 2015, and has been instrumental as the Chair of the Development Committee and supporting various research initiatives throughout his tenure.



Dr. Fluck worked at Franklin & Marshall College in the Biology Department for more than 30 years. He also served as an Associate Dean of the Faculty and was involved in establishing a partnership between F&M and CSC.

We thank Dr. Fluck for his tireless service to CSC and the countless contributions he's made over the years!

Stephen Tifft, MD

Dr. Tifft is joining the Clinic's Board of Directors in October. Dr. Tifft will be serving the remaining two (2) years of Dr. Stephen Ratcliffe's term. Dr. Ratcliffe joined our Clinic staff as a Senior Consulting Physician.



Dr. Tifft is the Managing Physician and a Pediatrician at Roseville Pediatrics, Lancaster General Health Physicians. Welcome, Dr. Tifft!

Glen Zimmerman

Mr. Zimmerman, of New Holland, PA, is joining the Clinic's Board of Directors in October. A native of Lancaster County, his school years were fulfilled at Farmersville Mennonite and Hillside Parochial School. His family business, Raytec LLC, provided many learning experiences and opportunities from a very early age which progressed into a partial ownership as it continues this day. His church affiliation is with Weaverland Mennonite Conference.



In July of 1995, Cristine Brubaker, became Cristine Zimmerman "as my beautiful and precious wife. We have been blessed with 7 children, 1 of which was born as a special needs (HPE) child. We had the joys and sorrows of 4.5 years of taking care of him, until he went Home to Eternal rest. This experience was our window into the unique organization of CSC. We feel indebted and grateful for their services and support and wish to be a contributor to the ongoing work and vision of CSC."

It's Flu Shot Season

Please call the Clinic at 717-687-9407 to schedule your flu shot. Anyone over six months of age is encouraged to get the flu shot. We offer shots to our patients and their immediate family 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.

2019 Extraordinary Give

On Friday, November 22nd, visit ExtraGive.org and select 'Clinic for Special Children' as your non-profit of choice! Last year over \$65,000 was raised for the Clinic in just 24 hours! Every dollar donated through ExtraGive.org will be stretched by a pool of over \$500,000.

If you would prefer to donate over the phone, please call the Clinic on November 22nd between the hours of 9 a.m. - 5 p.m. and we would be happy to receive your gift.

Visit us on 11/22 in Lancaster City's Penn Square from 11 a.m. - 3 p.m. for our annual Whoopie Pie Toss. We appreciate your support of the Clinic and the 2019 Extraordinary Give!



Photo credit: Eric Forberger

Thank you for helping!

Drs. Fox & Demczko

Last year Drs. Mike Fox and Matt Demczko served as members of the CSC clinical team coming one day a week each until an open full-time physician position was filled. With the hire of Dr. Laura Poskitt, Drs. Fox & Demczko will return to working solely at Nemours. We are so thankful to both Nemours and Drs. Fox and Demczko for their willingness to provide excellent care to CSC patients while we identified a new full-time physician.

Drs. Fox and Demczko had previously spent time at the Clinic for six month fellowships before they opened the Kinder Clinic in Dover, DE. The Kinder Clinic is owned and operated by Nemours and is the first of its kind to offer services directed toward children with special needs in southern Delaware from the Plain communities.

Thank you Drs. Fox & Demczko - your service has been invaluable and you will always be a part of the CSC family!

CSC Books | Purchase Options

If you'd like to purchase a copy of *30 Stories for 30 Years* or *Our Story in Newsletters*, please see the below purchase options available.

If you have any questions, please don't hesitate to contact us!

Mail a check to:

Clinic for Special Children
Attn: Kelly Cullen
535 Bunker Hill Road
Strasburg, PA 17579

If you'd like your book(s) shipped, please add \$3.50 per book. Please note on check memo or separate note which book(s) you're ordering.

Email

kcullen@ClinicforSpecialChildren.org

Call

717-687-9407

Purchase Online

www.ClinicforSpecialChildren.org/Store

Visit Us

at the Clinic for Special Children

30 Stories for 30 Years



\$50.00

The 30-year history of the Clinic for Special Children as told by first-hand accounts of those who've walked through its doors.

Read handwritten, first-hand stories and experiences of those who've bravely faced rare genetic diseases. Learn about how rare genetic diseases aren't so rare for those who have them.

And most of all, share compassion for those who aren't so different from yourself.

Order your copies today!

Our Story in Newsletters



\$50.00

From it's humble beginnings, the Clinic for Special Children has provided a medical home for children and adults with rare genetic diseases.

This book chronicles the 30-year history of the Clinic through its entire collection of newsletters.



Natural History Studies

What are they and why are they important?

When researching and evaluating the effectiveness of a new clinical treatment, there is a standard process researchers will follow. Typically, researchers will break up the patients into two groups. The first is called the “experimental” group and all patients in this group will receive the new treatment being evaluated. The second, called the “control” group, does not receive the treatment. Patients and researchers are typically “blinded” as to which patient is in what group, meaning neither knows if someone is receiving the actual treatment or a “placebo” which does not cause an effect. This process protects against bias. At the end of the study, the outcomes of the two groups can then be compared against each other to see if the new treatment creates a beneficial and meaningful difference.

However, this is rarely how we conduct studies at CSC. Since almost all patients seen at CSC have rare disorders, there are usually not enough people in the world with a given disorder to create the two groups of sufficient size. In addition, patients can have such severe disease that to not offer a promising treatment would be unethical. However, it is still important to be able to determine if the treatment is actually making a beneficial and meaningful difference.

Understanding the natural course of a given condition, without significant clinical intervention, is important because that information can be used as a “control” group when testing a new treatment. The process of gathering this information is called a natural history study.

Natural history studies are conducted in two ways. The first is a retrospective study. This is done by looking at historical patient data already available. This can be gathered from medical records, patient and parental interviews, family surveys, and other historical sources. In this type of study, data is collected by looking in the past.

The second type of natural history study is a prospective study. In this

approach, researchers identify specific measures to track over time and then carefully collect that data through various interactions with patients. These interactions can include lab testing, specialized exams, or imaging. The measures selected for this type of study are typically informed by information collected through a retrospective study.

At CSC, we are designing a number of prospective studies for conditions we think could be candidates for innovative treatments. This is of critical importance so that when the opportunity to treat presents, we will have the information available to quickly put a treatment study in place.

Recently, CSC conducted retrospective natural history studies on three conditions well-known to many of the families we serve. These disorders included Amish Nemaline Rod Myopathy (also called Chicken Breast Disease), which is caused by variants in the gene *TNNT1*, GM3 Synthase Deficiency which is caused by variants in the gene *ST3GAL5*, and Spinal Muscular Atrophy (SMA) which is caused by deletions in the gene *SMN1*. In each of these studies, medical records were scoured and parents of the patients provided important information about disease course, survival, hospitalizations, and other important milestones.

Specifically, the GM3 Synthase study was conducted as a collaborative effort with the members of the Plain Community Health Consortium (PCHC). Each study resulted in a peer-reviewed publication which shares this important information with researchers around the world.

We thank all the families who have participated or are currently participating in a natural history study at CSC. These studies are critical to enhancing our knowledge of all aspects of the course of specific diseases and our ability to identify and implement new and innovative treatments to the difficult problems faced by these families daily.



Collaborator Spotlight: Regeneron

Pictured above (L to R): Dr. Claudia Gonzaga-Jauregui and Dr. Cristopher Van Hout

As a graduate student at the University of Michigan, Dr. Cristopher Van Hout, Statistical Geneticist at Regeneron Genetics Center (RGC), read numerous publications by Dr. Erik G. Puffenberger, Laboratory Director at the Clinic for Special Children (CSC). The publications were useful as he studied human genetics and statistics and worked on his thesis focusing on the population genetics of the Old Order Amish of Lancaster, PA. Years later, as a collaborator of CSC, Dr. Van Hout's early learnings about the Clinic still hold true – that we understand the value of medicine informed by genetics at the community level.

When he learned about the scale of the human genetics research projects happening at RGC, he was “excited to join as the first member of a dedicated quantitative genetics group.” At the time, Dr. Van Hout didn't know that RGC would establish collaborations with organizations working with the Anabaptist communities, but he's enjoyed his career coming full circle.

Dr. Claudia Gonzaga-Jauregui was born and raised in Mexico City. In 2008, she moved to the United States to complete her PhD program in Molecular and Human Genetics at the Baylor College of Medicine in Houston, TX. During her studies she worked closely with the Human Genome Sequencing Center, specifically in projects that looked at genetic variation in human populations. She was also a part of the team that worked on the first proof-of-concept studies using whole genome sequencing and exome sequencing to identify causes of genetic disorders in patients with undiagnosed diseases.

In 2014, she was recruited to Regeneron to lead family-based analyses and collaborations at the newly created, RGC. The collaboration with the Clinic for Special Children was one of the first that Dr. Gonzaga-Jauregui worked on at her new job. She immediately began performing genetic analyses of Amish and Mennonite families and children with suspected genetic disorders. As this was her first interaction with the Plain communities, she dove into learning more about the population history and what was already known about the genetics of the Plain people. This research and learning allowed her to perform better genetic analyses of the families that were referred from CSC.

CSC uses a standard process to attempt to determine the genetic cause of disease in new patients. First, the patient is examined by one of CSC's clinicians to determine if the problems the patient is experiencing is likely to have a genetic cause. If it looks like the issue is related to genetics, then CSC's lab may check for variants that have already been identified. If everything already known seems unlikely or the tests are negative, the CSC's lab will run a test called a microarray. This looks for regions where there may be too many copies or not enough copies of DNA. An example of a disorder that could be diagnosed with a microarray is Down Syndrome, where a patient has three copies of Chromosome 21 rather than the typical two.

If the cause of disease is not able to be determined at this point, then CSC turns to RGC. Blood samples are collected from the patient as well as their parents and siblings and DNA is sent to RGC for exome sequencing. Exome sequencing is an advanced molecular test where the 1-1.5% of a person's DNA that serves as the blueprint for the production of protein is sequenced. This process yields the exact chemical make up of the DNA.

However, sequencing the DNA is just the start of the process. Each time someone's DNA is exome sequenced a total of about 70,000 genetic changes are identified. Researchers at RGC and CSC compare the changes to various databases and the results from close family members to determine which change or changes are causing the patient's clinical issues. Sometimes the changes are inherited from the parents. Sometimes a change is spontaneous and is only present in the patient and no other family members. These are called *de novo* variants because they are “new” and not inherited. Sometimes the test does not reveal an obvious solution. After the analysis is complete, the CSC team will share the results with the family and counsel them on their implication.

To date, the RGC team has sequenced and analyzed about 250 Amish and Mennonite families in collaboration with CSC. In most of those cases, a genetic cause of disease was determined. Sometimes the cause was in a gene known to cause disease. While in other cases the cause was in a gene not known to cause disease and had never been seen before. We are so thankful to the RGC team for their continued collaboration and helping us provide answers to many of our patient families!

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
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Yalonda L. Kosek
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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
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William Van Ess, MS, CFE
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Millie Young, RNC
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Donor Spotlight

Lori Hartmann-Borris | WeeUsables

As a local mom Lori Hartmann-Borris, owner of WeeUsables Consignment Events, knows that saving on children's items and turning your children's clutter into cash is the best! That's why she created WeeUsables Consignment Events, to bring together local families in Lancaster, York, and Harrisburg to consign and purchase children's clothes, toys, and more.

In 2009, Lori purchased the former Finders Keepers for Kids sale and has since re-named, expanded, and reimagined the consignment events. She held the first WeeUsables event in the spring of 2010 at the Lancaster Farm and Home Center. Since that first sale, two more locations have been added including a new Harrisburg sale this year that takes place on November 8-9th at Sports City Athletics. To date, WeeUsables has officially had over 1,200 consigning families, 250,000+ kid's items, 8,000+ happy shoppers, and \$92,000 donated to local charities. The Lancaster sales typically have inventory that exceeds 130,000 items and shoppers can save up to 90% off of retail prices. Each sale starts with a pre-sale for first time parents, grandparents and foster parents as well as a public pre-sale with pre-purchased tickets. After the pre-sale period, the public sale is open to all with free admission.

At the end of each WeeUsables sale, there is a 'Stuff-A-Bag' charity event where any leftover items are for sale for \$10 a bag. Shoppers can fill up their bags with merchandise and all of the proceeds benefit a local charity. For the 2019 Lancaster Fall sale, the Clinic for Special Children was chosen as the benefiting charity! In addition to the Stuff-a-Bag event, the WeeUsables team accepted donations to the Clinic at each of the checkout registers during the event. The Clinic team also had a table throughout the event with merchandise for sale and informational brochures.

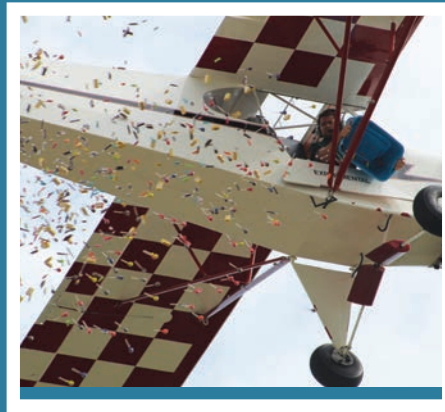
The 2019 Fall Lancaster sale ran from September 4-7th at Spooky Nook Sports and was one of the biggest sales yet for the WeeUsables team. After the sale, WeeUsables donated almost \$4,500 to the Clinic for Special Children! We are so thankful to Lori, and all of the consignors and shoppers at the Lancaster sale!



2019 BENEFIT AUCTION SEASON

To everyone that attended an auction, donated an item, volunteered their time, or supported in any other way – THANK YOU!

Our 2019 benefit auction season was a record-breaking one and we're happy to look back on this auction season with the selected photos below.





Clinic for Special Children

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ClinicforSpecialChildren.org

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The Clinic for Special Children is a Section 501(c)(3) Public Charity for US Federal and State Tax purposes (Tax ID #23-2555373). Donations to the Clinic are tax deductible. Donors should consult their tax advisor for questions regarding deductibility. A copy of the Clinic's registration and financial information may be obtained from the PA Department of State by calling toll free, 1-800-732-0999.

2019 ExtraGive

Think of the Clinic for Special Children on
Friday, November 22nd - Lancaster's largest day of
online giving!

Last year we raised over \$65,000 in less than 24 hours!
Will you help us reach our **goal of \$70,000 this year?**

See the insert and page 2 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 consultive services, and by increasing and disseminating knowledge of science and medicine."