2018

ANNUAL REPORT

Clinic *for* Special Children

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Our 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.

Our Vision

We envision the Clinic for Special Children as a MEDICAL HOME

for predominately Amish and Mennonite children and adults who are born with genetic predispositions to disability, chronic disease or untimely death. We continually strive to integrate advanced scientific tools and concepts into clinical practice so that genetically vulnerable people have access to the most timely, affordable, and effective healthcare. The Clinic for Special Children represents an innovative and holistic approach to modern medical care that can inform the practice of genomic medicine in other settings. We seek opportunities for education and collaboration that promote the well-being of genetically disadvantaged, underserved individuals throughout the world, and are dedicated to training the young clinicians and scientists who will care for these individuals now and into the future.

GENERAL *STATS*

1,070 *active patients*

18 *staff members*

Advancing Science and Medicine, One Child at a Time.

Friends,

As we reflect on the past year at the Clinic for Special Children, we feel a deep sense of gratitude. We welcomed new staff members, created new community events and initiatives, built valuable partnerships, and advanced research for the treatment of rare genetic diseases.

In every facet of our organization, 2018 has proven a banner year. On September 22, 2018, we hosted our first annual Clinic for Special Children 5k, welcoming more than 300 participants and raising more than $24,000. Thanks to donors like you, we had record-breaking fundraising results this year. During the Extraordinary Give in November, Lancaster County's largest day of online giving, more than $65,000 was raised for the Clinic in just 24 hours!

A new Research Operations team was established in January to ensure patient-focused research and translational medicine is always at the forefront of our everyday work. An advanced next-generation sequencing strategy, the Plain Insight Panel, was designed and tested to include 1,300 genetic variants that are linked to health problems within Plain populations. This test will help us identify couples who are at risk for having a child with a treatable genetic disease to allow for timely diagnosis and the highest quality of preventative care.

As we look ahead to 2019, we will be commemorating the Clinic's 30th anniversary – a milestone that couldn't have been achieved without your support. On behalf of our staff and board, we thank everyone who has visited the Clinic, shared in its mission, participated in community fundraisers, or otherwise supported work on behalf of the families we serve.

Adam D. Heaps, MS, MBA
Executive Director

Kevin A. Strauss, MD
Medical Director

Patient Care

Delivering effective and

AFFORDABLE CARE *for* INDIVIDUALS

with genetic conditions.

PATIENT CARE *STATS*

5,008 *biochemical & genetic tests*

1,744 *patient visits*

Manage **306** *known variants that cause disease*

42 *states &* **17** *countries*

"None of this work or inspiration can be accomplished by any one person. To me, the Clinic has always been more about an idea than any individual or group. And it's the kind of idea that we all share."

- Dr. Kevin A. Strauss, *Medical Director*

Patient Focus

"AN ANGEL AMONG US", is how Ruth Martin describes her son, Darren Lee Martin ('Lee'). Pictured to the right with his parents, Ruth and Darren, and older brothers Adam and Tristan, Lee's joy is evident.

Born in a hospital setting, there was nothing notable about Lee's birth. But at six weeks old, mom and dad started to notice that Lee wouldn't giggle, laugh, and was unable to follow them with his eyes. After several visits to pediatricians and specialists, the Martin family was referred to Clinic for Special Children (CSC).

Lee first visited CSC when he was six months old. Dr. Strauss worked to manage Lee's symptoms while Dr. Puffenberger worked towards a genetic diagnosis. When CSC started collaborating with the Regeneron Genetic Center in 2014, the Martin's DNA was one of the first sent for an advanced genetic testing called exome sequencing. A mutation was found in one of the five genes known to cause

Kleefstra Sundome (KS). Like most children with KS, Lee had a de novo mutation, meaning the genetic change causing his symptoms was not inherited from either parent. Kleefstra Syndrome is characterized by intellectual disability and a spectrum of physical and clinical symptoms. Since the symptoms can be diverse, it makes managing care challenging. Although there are no cures for KS, there are many options for the management of symptoms.

"Having a special child has changed our outlook on life. Most importantly, we are committed to live life as normal as possible and allow Lee to experience the same joys as other children." Since there is no definitive prognosis for Lee, the Martins face one day at a time. Darren and Ruth have found strength in their faith and support from their family and friends.

Now a vibrant nine-year-old boy, Lee's favorite things are to wrestle with his big brothers, spend time outdoors, take car rides, and swinging. "We are so grateful to Dr. Strauss and the Clinic team for their compassion. Dr. Strauss has made a big difference for us, always making sure we, his family, are also taken care of as well as Lee," says Ruth.

"Life doesn't stop when your child is diagnosed with a rare disorder." To parents living with special children, Ruth offers the advice, "Through the roller coaster of emotions you may experience, give yourself time. Wherever you are in your journey, it's OK to be where you are and feel the way you do. Everybody's story is different and we wanted to tell ours in case we could inspire one person."

Front (L-R): Adam and Darren Lee Weaver
Back (L-R): Ruth, Darren, and Tristan Weaver

Outreach and Community Empowerment

COMMUNITY

The first annual Clinic for Special Children 5k was held on Saturday, September 22, 2018 on a bright, sunny Fall morning. Over 280 community members came to the Clinic to run, jog or walk, and over $24,000 was raised to benefit the Clinic! The Clinic staff, sponsors, volunteers and participants came together to support the Clinic in providing comprehensive clinical, laboratory and diagnostic services for children living with rare genetic diseases.

The day included a silent auction which featured beautiful quilts, tickets to Sight & Sound Theatres and Strasburg Rail Road, gift cards to local grocery stores, eateries and more! The team from Zolē Art Factory in Strasburg offered paint-your-own children's crafts to spark some creativity in the little ones. Members from the Plain community organized a bake sale, which included apple dumplings, delicious pies, whoopie pies, cookies and breads.

Thank you to everyone who made the day a successful one. We are looking forward to our 5k on September 21, 2019!

OUTREACH

While many families often travel considerable distance to receive medical care, four-year-old Margaretha and her parents made a nearly 2,500 mile journey from southeast Mexico to Delaware to do so. They are Old Colony Mennonites who live in the Campeche region of Mexico, descended from Prussian Mennonite colonists who migrated from Russia to settlements in Canada in the 1870s, and then into Mexico a few generations later. Dr. Strauss and Karlla Brigatti traveled to the colony last August with CSC Board Chairman, Herman Bontrager, members from Mexico Mennonite Aid (MMA), and the Anabaptist Foundation on a medical mission. Several families there have children with special needs so Dr. Strauss and Karlla met with over 30 patients for several days and devised care plans that could be implemented locally. However, some children had serious medical problems that could not be managed in Mexico, including Margaretha. She was born with cloacal extrophy, a very rare and severe birth defect where most of the abdominal organs, including the bladder, intestines, and spinal cord, are externalized and exposed early during development in the womb, causing multiple serious health issues after birth. Margaretha was able to have some of the necessary surgeries in Mexico, but she still required constant care and remained at risk for life-threatening complications. Despite these challenges, she was a lively, happy child, and her family was very dedicated to her care.

Once back home, Dr. Strauss worked closely with International Medicine at the Nemours/ Alfred I. duPont Hospital for Children, the Anabaptist Foundation, and MMA to determine the feasibility and logistics for Margaretha to have surgery at Nemours. At long last, Margaretha and her parents were able to come to Nemours in early June 2018. Her surgeries took over 14 hours, but she made a full recovery and will have a much better quality of life as a result. She also enjoyed her stay at the Ronald McDonald House at Nemours and the family found everyone at the hospital incredibly helpful and kind. When Dr. Strauss and Karlla went to Nemours to see her in late July, Margaretha's parents were looking forward to returning to Mexico and resuming their normal lives, but happy they made the trip and very pleased with Margaretha's care. The joint efforts from the Clinic for Special Children, MMA, the Anabaptist Foundation, and Nemours made a big impact on a very special little girl!

EDUCATION & COMMUNITY *STATS*

7 *family days in 2018*

15 *research students*

Research

This year, 7 Clinic staff members contributed to original research published in

JOURNAL OF PEDIATRIC ORTHOPAEDICS

The report identified an alternative method to deliver nusinersen to patients with Spinal Muscular Atrophy (SMA) using a subcutaneous intrathecal catheter system (SIC) configured by connecting an intrathecal catheter to an implantable infusion port. SMA is a devastating genetic disease that leads to progressive degeneration of motor neurons that control movement, swallowing, and breathing. It is the leading genetic cause of infant death worldwide. Nusinersen is the first FDA approved therapy for SMA but must be administered into the cerebrospinal fluid by repeat lumbar puncture every 4 months of life. Unfortunately, the majority of surviving SMA patients have skeletal deformities or spinal hardware that make it difficult to safely and reliably access the cerebrospinal fluid. Ten SMA patients underwent implantation of the catheter devise and received nusinersen dosing through the SIC.

READ THE ARTICLE ONLINE AT:

https://bit.ly/2wBOHFc

RESEARCH & DEVELOPMENT *STATS*

42 **NEW** *disease-causing genetic variants*

6 *peer-reviewed publications*

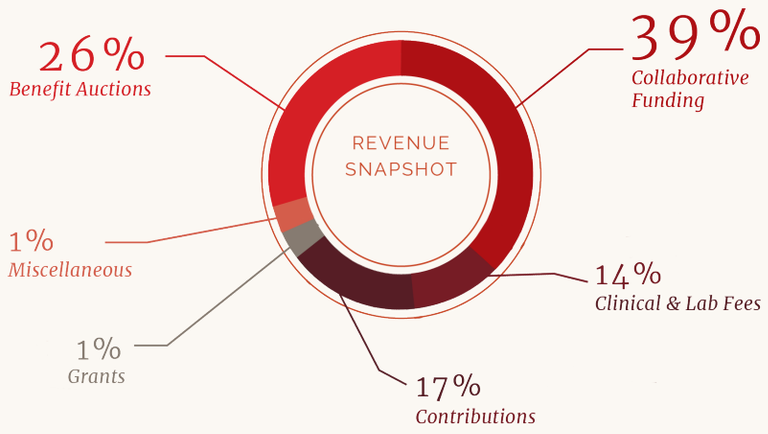
Donor Spotlight

Strasburg Rail Road

Let's travel back in time to the early 1800's in Lancaster County - the Philadelphia and Columbia Rail Road was one of the earliest in the area, and unfortunately bypassed the town of Strasburg by a distance of four to five miles. The townspeople of Strasburg petitioned to have a short line railroad connect to the Philadelphia-Columbia Rail Road and in 1832, the Strasburg Rail Road was chartered by PA legistlature. From the beginning of the operation of the Strasburg Rail Road until 1958, there was an ebb and flow of good times and bad. Following WWII, the construction of interstate and major highways threatened the way of rail roads and left the Strasburg Rail Road eventually in a state of near abandonment. Upon hearing of the dismal fate of the Strasburg Rail Road, a group of twenty-four "rail fans" in Lancaster County bought the property and rail line. On a whim, they decided to buy an old wooden coach, remodeled it with a tiny gasoline engine, and started to take small groups of people on train rides through the Lancaster countryside. It was a successful combination of history, nostalgia, Amish culture, and Lancaster County countryside that garnered the rail road such interest from the public. Today, the Strasburg Rail Road is working on their 20th passenger car to come out of the shop this winter.

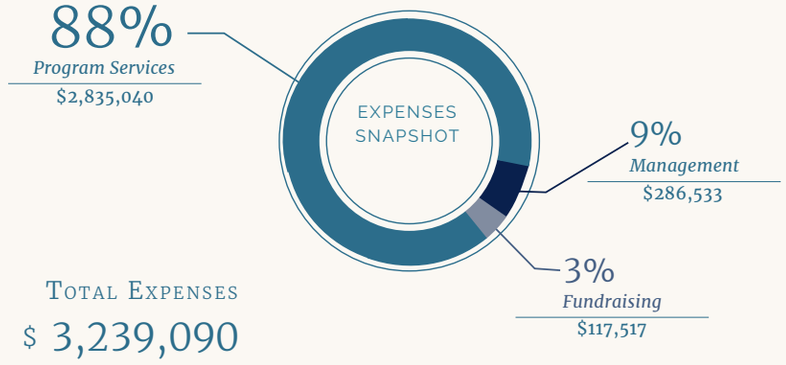
Several years ago, the Strasburg Rail Road chose the Clinic for Special Children as their "charity of choice" to donate funds from their Great Train Robbery and Christmas events. Steve Barrell, Station-master at the Strasburg Rail Road explained, "We are glad to support and partner with an organization right here in Lancaster County that has a local mission and works with children and families." Since the initial partnership, the Strasburg Rail Road has donated over $36,000 to the Clinic for Special Children and has made thousands of visitors to Lancaster County aware of the Clinic's mission and history. We are so thankful for the partnership with the Strasburg Rail Road and are excited to partner on future events together!

2018 Revenue Snapshot



The Clinic is funded from four major sources of revenue—annual auctions, donations from caring people just like you, collaborative relationships, and fees for services paid by patient families.

Your support enables CSC to continue to provide patient with timely, affordable, and effective care! We work hard to keep clinic fees to a minimum and effectively utilize funds to fulfill CSC's mission.



2018 | FINANCIALS

STATEMENT OF FINANCIAL POSITION
As of 9/30/2018

ASSETS

Cash and Equivalents	$1,207,794
Accounts Receivable	$118,286
Promises to Give	$0
Prepaid Expenses	$8,692
Property and Equipment	$678,501
Investments	$3,731,417
Investments Restricted	$953,921
Total Assets	$6,698,611

LIABILITIES & NET ASSETS

LIABILITIES	
Accounts Payable	$217,169
Accrued Expenses	$25,162
Accrued Wages	$29,616
Deferred Revenue	$6,608
Total Liabilities	$278,555
NET ASSETS	
Undesignated	$1,733,672
Board Designated	$3,706,897
Temporarily Restricted	$699,977
Permanently Restricted	$279,510
Total Net Assets	$6,420,056
Total Liabilities and Net Assets	$6,698,611

STATEMENT OF ACTIVITIES
10/1/2017-9/30/2018

REVENUE

Contributions	$636,892
Grants	$40,000
Collaboration Funds	$1,337,565
Special Events	$905,713
Clinic Fees	$306,524
Investment Income	$361,852
Laboratory Fees	$177,256
In-Kind Donations	$93,000
Miscellaneous income	$24,220
Total Revenue	$3,883,022

EXPENSES & PROGRAM INVESTMENTS

Program Services	$2,835,040
Management	$286,533
Fundraising	$117,517
Total Expenses	$3,239,090

Front (L-R): Ashlin Rodrigues, KaLynn Loeven, Emily Seitz, Kevin Strauss, Karlla Brigatti, Donna Robinson, Candace Kendig, Lauren Bowser

Back (L-R): Erik Puffenberger, Christine Hendrickson, Yalonda Kosek, Millie Young, Vincent Carson, Keturah Beiler, Teresa Rineer, Julia Martin, Kelly Cullen, Adam Heaps (Not Pictured: Lavina King)

Clinic *for* Special Children

Our Staff

Keturah Beiler, RN
Nurse

Lauren E. Bowser
Research Fellow

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

Vincent Carson, MD
Pediatric Neurologist

Kelly Cullen
Communications Manager

Adam D. Heaps, MS, MBA
Executive Director

Christine Hendrickson, RNC
Nurse

Candace Kendig
Medical Receptionist

Lavina King
Community Liaison

Yalonda L. Kosek
Office Coordinator

KaLynn Loeven
Laboratory Technician

Julia Martin
Development Assistant

Erik G. Puffenberger, PhD
Laboratory Director

Donna L. Robinson, CRNP
Nurse Practitioner

Ashlin Rodrigues
Laboratory Technician

Emily Seitz
Scientific Grant Writer

Kevin A. Strauss, MD
Medical Director

Millie Young, RNC
Research Nurse

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Herman Bontrager
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Richard Fluck, PhD
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Chair-Development Committee

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