

Meaningful Momentum

2021
Annual
Report



Clinic for
Special Children®

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

COMPREHENSIVE MEDICAL SERVICE

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 timely, affordable, and effective healthcare. The Clinic 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

32 *years serving the community*

26 *staff members*



Meaningful Momentum

Pictured above (L-R): Adam Heaps, MS, MBA,
Kevin A. Strauss, MD, and Herman Bontrager

Friends,

Reflecting back on our work this past fiscal year, we see meaningful momentum in all areas of the Clinic for Special Children's mission. Our continued patient-driven growth and life-changing work is only possible through your steadfast support. We are deeply grateful for each of you.

While the past few years have not been without challenges, we have continued providing care without interruption. Your support ensures our clinicians can always attend to the needs of children and families, and provide them with the reassurance that comes from knowing we'll be there when they need us most.

In our day-to-day work, we continue to see a growing demand for our clinical and laboratory services as a direct result of major advances made by the Clinic over our three-decade history. Many children with previously fatal genetic disorders are now surviving and thriving into adulthood. Carrier screening and genetic testing in the community

continues to grow as word spreads about the vital importance of preventative medicine.

Alongside clinical care, the Clinic has an active, patient-focused research agenda. Our clinicians and scientists partner closely with patients and collaborators in the quest to find new treatments for some of the most prevalent and challenging genetic disorders that burden the Plain community. In service of this mission, our research team published 11 peer-reviewed publications this past year, sharing critical information with the larger medical community. In addition, we engage in clinical trial work to explore and expand access to new therapies for the children and adults we serve.

As we begin our 33rd year, we are enthusiastic and hopeful about the work that lies ahead. Our interdisciplinary team collaborates every day to keep the promise of our mission of providing care to people living with rare diseases, continuing the search for vital treatments, and serving the communities that entrust us with their care.

Adam D. Heaps, MS, MBA
Executive Director

Kevin A. Strauss, MD
Medical Director

Herman Bontrager
Chair of the Board

Caring Today & Hoping for Tomorrow

Treating & researching TNNT1 Myopathy

JOSIE'S STORY

The Clinic for Special Children has a unique care model where clinicians, caregivers, researchers, and scientists work hand-in-hand to solve complex medical and genetic questions. Michael and Esther Stoltzfus witnessed the embodiment of this commitment of working together though the care provided to their son, Josie.

Shortly after Josie's birth, his parents noticed that he started to have tremors. The Stoltzfus family quickly brought Josie into the Clinic for Special Children for care. On the day of his first visit to the Clinic, Josie was diagnosed with TNNT1 myopathy, a devastating rare genetic disorder that affects the muscles.

Josie's family and clinicians worked together to develop a care plan and enrolled him into the Clinic's home-based, free of charge palliative care program, Cherished Lives. The ability to have compassionate care provided in their home on a weekly basis through Cherished Lives was priceless to the family. "I don't know where we would have been without the Clinic," Michael says, "Anytime we needed something, the Clinic was there."

Alongside Josie's care through Cherished Lives, the Stoltzfus family worked with the Clinic's team on research for TNNT1 myopathy. Josie was enrolled in the Clinic's WiTNness research study, a natural history

study of TNNT1 myopathy. "We decided to do the research study so Josie's life could help others. It also gave us fresh hope for a treatment one day soon," says Michael.

Sadly, Josie died in the summer of 2020 at 14 months of age. "We want to share Josie's story in the hopes that it can help others. He brought so much joy to our lives," says the Stoltzfus family, "It's a different kind of love for a special child - it makes you focus on being present and take one day at a time."

Josie's story, and countless of others, reaffirms the dedication of the Clinic team in the search of treatments for rare genetic disorders. The WiTNness study aims to get the Clinic ready for a future clinical trial of a potential treatment for TNNT1 myopathy, like gene replacement therapy. "It gives us the baseline we need, measures we can use and look for change when we give a therapy, and allows us to measure that therapeutic response in an informative and consistent way," explains Karlla Brigatti, MS, CGC, Research Operations Director at the Clinic.

TNNT1 myopathy demonstrates how clinical care, research, and laboratory services at the Clinic work together for a common goal. For the Clinic, the most important goal is to help patients with genetic conditions overcome limitations in treatment options and provide them with better outcomes and quality of life.

PATIENT CARE STATS

1,252 *active patients*

from **44** *states &* **17** *countries*

5,652 *laboratory tests to help identify & manage genetic illnesses*

1,260 *patient visits*



A Double Diagnosis

Treating & caring for SMA & MSUD

MADELYN'S STORY

Wilson and Elva Martin received surprising news a few days after their daughter Madelyn's birth. She was the first baby known to be diagnosed with the two rare genetic disorders Spinal Muscular Atrophy (SMA) and Maple Syrup Urine Disease (MSUD). "It was a blow, but not a shock. We had previously done carrier testing and knew what we were carriers for, but were surprised to get two diagnoses," says Elva Martin. After receiving the diagnoses, the family had to decide where to go for Madelyn's unique care.

The Martin's had first learned about the Clinic for Special Children (CSC) after attending a benefit auction in Missouri. They were already a part of the rare disease community, as their firstborn son was also diagnosed with SMA. After Madelyn's diagnoses, the family faced an important question: should they care for Madelyn at a larger health system close to home, or drive to Pennsylvania to seek care with CSC? "We decided to care for Madelyn in Pennsylvania with the CSC team where they could manage her genetic disorders and they were very familiar with the process of administering SMA gene therapy," explained Wilson and Elva. The Janet C. Scala Patient Access Fund at CSC helped offset the family's cost of cross-country travel and long-term lodging in Lancaster County.

After the family decided to come to PA to seek care, the CSC clinical team jumped into action to develop a care plan with the family. With multiple treatments available for SMA, the family was presented with all options and ultimately decided upon targeted gene therapy.

The CSC's expertise in working collaboratively to administer disease-modifying treatments, like gene therapy, was one of the main reasons the family entrusted their care at CSC.

Upon arriving in PA, the CSC team collaborated with Penn Medicine Lancaster General Hospital to have SMA gene therapy available for the family. While Madelyn's MSUD amino acid levels were high upon admittance to the hospital, the CSC clinical team was able to stabilize her MSUD levels and administer her the SMA gene therapy at just 12 days old.

Madelyn continues to receive compassionate and coordinated care at CSC with regular check-ups every three to five months. Due to CSC's decades of innovation in the care of MSUD, the family sends in filter paper tests twice a week to monitor Madelyn's amino acid levels to ensure they don't reach dangerous levels. "The CSC team is definitely very experienced and we feel comfortable having our children in their care. They're a great bunch of friendly people and we feel at home at CSC. I'm just glad we go to CSC, it's a great place," says Wilson and Elva.

Just several years ago before the innovation of today's SMA treatments, babies diagnosed with SMA type 1 typically did not reach any milestones and had a life expectancy of around six months. Today, Madelyn is a year old and reaching milestones like sitting and (almost!) crawling! The care that Madelyn receives represents the cornerstone of the work at CSC - providing the right treatment, for the right child, at the right time through coordinated, compassionate, and personalized care.



RESEARCH STATS

21 *NEW disease-causing
genetic variants identified*

11 *peer-reviewed
publications*

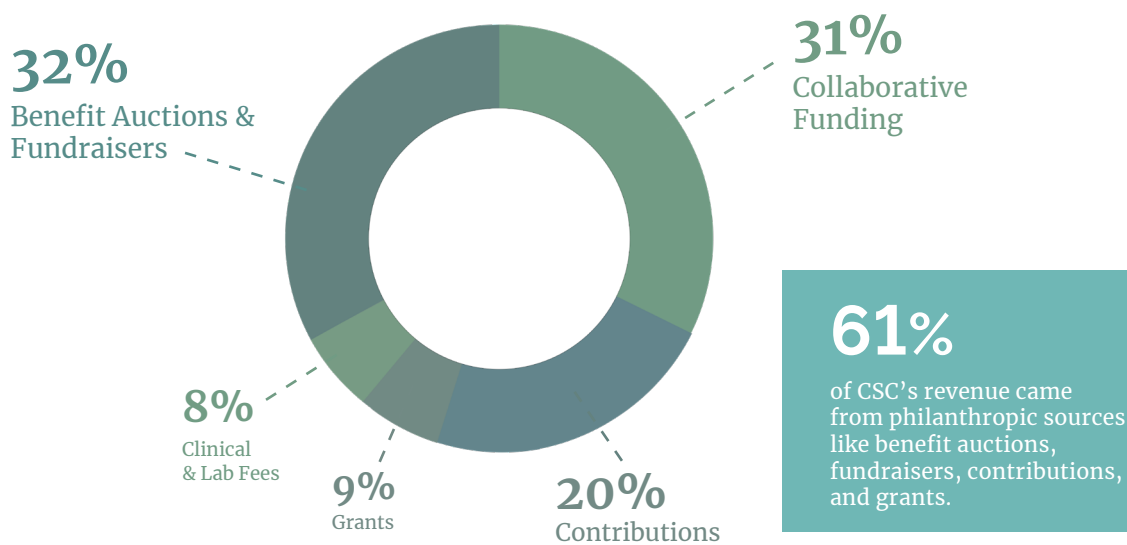
Financials Overview

A fiscal year in review

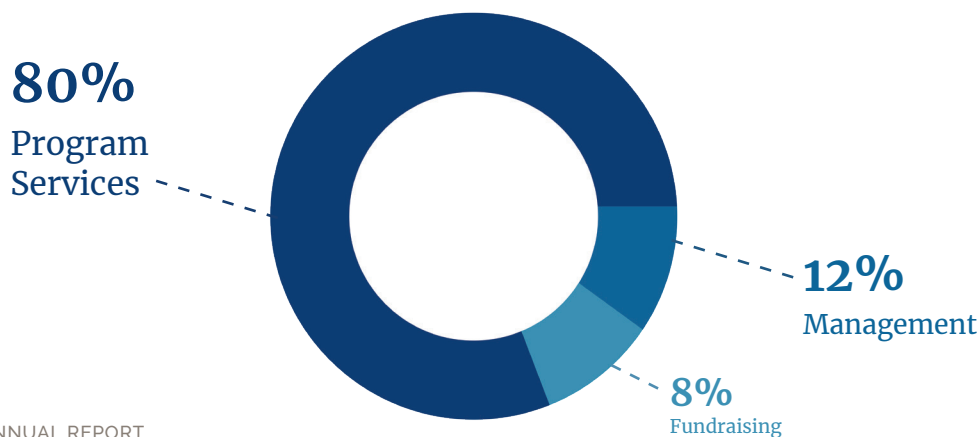
Only about 10% of the Clinic’s annual operating budget is derived from patient and laboratory fees. In order to provide accessible care, our clinical and laboratory fees have not been raised in over a decade. The remaining 90% of the funds needed to provide timely, affordable, and effective care come from philanthropic sources like benefit auctions, fundraisers, contributions, and grants, as well as collaborative relationships.

As a 501(c)3 non-profit organization, philanthropy and support from the community is vital to achieving our mission. Your support enables us to continue caring for special children and adults.

2021 REVENUE SNAPSHOT



2021 EXPENSES SNAPSHOT



Planning for the Future

Strategic planning & building on strong ground

In just a matter of a decade, the Clinic for Special Children has experienced unprecedented growth. Just 10 years ago we had 10 staff members, and today, we now have 27 team members working to fulfill our mission every day. Our waiting rooms are often at capacity, parking lots are full, and previous conference areas have been converted into office space for our growing staff. To address this growing issue, we purchased a 10.27 acre plot of land in Leacock Township, Lancaster County to plan for building growth in the near future. We look forward to sharing more details and plans with you on this project as they become available.



In order to ensure that we are building on strong ground, we started a new strategic planning process this year. We invited our entire staff, board, and key

stakeholders to contribute to the planning process to ensure that every strategic area of the Clinic was represented. We are working through collecting the feedback, prioritizing key initiatives, and developing clear goals for the Clinic to outline our future plans.

While we are still in the process of developing a strategic plan for the next five years, we are looking forward to sharing the details with you later in 2022. It's been an invigorating and energizing experience to envision what the next five years and longer can hold for the Clinic for Special Children. The one thing that we know now and in the future without exception is that we cannot do this vital work without your continued support.

Thank you for cherishing the lives of special children and adults, and supporting us on providing affordable, effective, and timely care now and in the future.

FINANCIAL STATS

8%

*of revenue came from
patient & lab fees*

\$4.3 million
2021 operating budget

Genetics and the Plain Community, or, why so many Stoltzfuses?

An interview with Dr. Erik G. Puffenberger

Dr. Puffenberger completed his PhD in 1996 under the direction of Dr. Aravinda Chakravarti at Case Western University and joined the Clinic for Special Children in 1998, a time when the link between a person's genetics and their physical health was just beginning to be understood through studies like the Human Genome Project.

Over the past 24 years, Dr. Puffenberger has built the Clinic's laboratory from the ground up, played a central role in CSC's ability to identify novel, rare disease genes, developed innovative strategies for newborn and carrier screening in the Plain communities of North America, and made significant contributions to research on rare disease and public health.



Dr. Erik G. Puffenberger

In your experience of speaking to a broad range of audiences, from scientific colleagues at international conferences to Plain community members interested in managing their genetic health, is there a common question you tend to receive about your work?

Definitely. Most people are very curious about why certain rare diseases, for instance Maple Syrup Urine Disease or Glutaric Acidemia Type 1, are so common in the Plain communities we serve at CSC. The answer to this is complicated by scientific and cultural factors; however, we can point to a central genetic theory to help us understand the answer – random genetic drift.

What is *random genetic drift*, and can you tell us more about what it looks like in the Plain communities?

The term *random genetic drift* describes the phenomenon of random fluctuations in the frequency of a gene variant, or mutation, in a certain population. By chance, individuals inherit a mix of gene variants from each parent. Across generations, some gene variants can be inherited more often by chance, leading to a larger number of carriers, and an increased risk for the genetic disorder associated with that variant. On the flip side, some genetic variants may not be inherited often by chance, and their frequency in the population diminishes over time. This change in frequency of certain genetic variants is completely due to chance events, hence the name of this phenomenon – random genetic drift.

What does this look like in the Plain community? Based on the theory of random genetic drift, we believe that when Anabaptist (Amish and Mennonite) settlers first came to North America, one person carried the gene mutation for each rare genetic disease found in the Plain populations. As these small founder populations grew, genetic variants harbored by the founding individuals ei-

ther increased or decreased in frequency based on the number of times the variant was passed on to the children of carriers.

We have to think in terms of generations, as well as consider the role of culture, to get at the root of this question. Some rare diseases have become particularly prevalent in Plain communities because of random genetic drift and the practice of endogamy, i.e. the custom of looking within, rather than outside of, the community for marriage partners. Over many generations, a relatively small set of randomly-inherited genetic variants becomes more frequent in the population, leading to a higher rate of rare diseases like Maple Syrup Urine Disease than in the general (or outbred) population. On the other hand, some genetic variants decrease in frequency or completely disappear from the population when they are not passed down from generation to generation. For instance, Cystic Fibrosis, the most common pediatric genetic disease in people of European descent, is virtually absent from our local Plain communities.

Speaking of culture, can you tell us why there are so many Stoltzfuses in Lancaster County?

Stoltzfus has become the most prevalent Amish surname among the Lancaster County Amish, accounting for approximately 25% of Amish households, but it was not always so common. When the original settlers came to the United States in the 1700s, there was only one male Stoltzfus founder. Through multiple generations, the Stoltzfus surname has increased in frequency by chance. We can imagine many ways this might have occurred: perhaps more boys were born into Stoltzfus families, or maybe Stoltzfus families were larger compared to other Amish families. Whatever the underlying reason, the effect was to increase the number of men who would then pass along their surname to future generations. The same phenomenon can be seen with the Martin surname in Lancaster County Mennonite communities, which accounts for about 20% of Mennonite households. The prevalence of Stoltzfuses and Martins in the Plain communities of Lancaster County is a simplified way for us to think about how genetic drift works in a culturally closed community.

To read more about Dr. Puffenberger's work, please see his recent article in the American Journal of Medical Genetics, "Mendelian disease research in the Plain populations of Lancaster County, Pennsylvania" (2021).





Clinic for Special Children®

Our Staff

Clinical Services

Amy Albright, MS, CGC
Genetic Counselor

Keturah Beiler, BSN, RN
Nurse & Cherished Lives Program Manager

Vincent Carson, MD
Clinical Operations Director

Christine Hendrickson, RNC
Nurse

Alexis McVey, BSN, RN, CPN
Nurse

Grace Loudon Meier, MD
Family Medicine Physician

Laura Poskitt, DO
Pediatrician

Stephen D. Ratcliffe, MD, MSPH
Senior Consulting Physician

Donna L. Robinson, MSN, CRNP
Nurse Practitioner

Kevin A. Strauss, MD
Medical Director

Susan Walther, MS, CGC
Genetic Counselor

Research Operations

Emilienne Bolettieri
Research Associate

Karlla W. Brigatti, MS, CGC
Research Operations Director

Millie Young, BSN, RNC
Research Nurse

Laboratory

KaLynn Loeven
Laboratory Scientist

Erik G. Puffenberger, PhD
Laboratory Director

Ashlin Rodrigues
Laboratory Scientist

Development

Skye Gawn
Development Associate - Operations

Julia Martin
Development Associate

Emily Seitz, PhD
Development Director

Administration

Kelly Cullen
Communications Manager

Jennifer Giaccio
Medical Receptionist

Adam D. Heaps, MS, MBA
Executive Director

Candace Kendig, RMA
Office Manager

Lavina King
Community Liaison

Dawn Sheets
Medical Receptionist

William Van Ess, MS, CFE
Accounting Manager

Board of Directors

Jan L. Bergen

Cindy Bo, MBA
Secretary

Herman Bontrager
Chair of the Board

Peter Crino, MD, PhD

Elam Esh

Leon Hoover
Vice Chair

Leonard Hurst

Mark Martin
Treasurer
Chair- Finance Committee

Jacob Petersheim

Stephen Tifft, MD

Glen Zimmerman
Chair- Development Committee

535 Bunker Hill Road
PO Box 128
Strasburg, PA 17579

tel (717) 687-9407

fax (717) 687-9237

ClinicforSpecialChildren.org

The Clinic for Special Children is a Pennsylvania non-profit corporation and a 501(c)3 public charity for US federal and state tax purposes (Tax ID # 23-2555373). The official registration and financial information of The Clinic for Special Children, Inc. may be obtained from the Pennsylvania Department of State by calling toll free, within Pennsylvania, 1 (800) 732-0999. Registration does not imply endorsement.