Creating **Brighter Futures**

2023 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 PRACTICE

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 people 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*

34 *years serving the community*

31 *staff members*

Staff at the new facility | August 2023

Creating **Brighter Futures**

Friends,

For nearly 35 years, the Clinic for Special Children has stayed focused on its core mission: improving the quality of life for children and adults with genetic or other complex disorders. This mission would not be possible without the trust and partnership between the Clinic staff and the families served by our organization. While the specifics of how and who is advancing the mission have changed, our commitment to providing innovative but practical solutions to challenging medical questions has not wavered.

If you have been following the progress of our new building project, you will know a big change is coming in April 2024. After many years of consideration, planning, fundraising, and construction, we will move from Strasburg to Gordonville, PA and settle into a new, expanded, and updated facility.

This project was pursued to ensure the Clinic has the resources needed to continue to fulfill our mission for many more years. We're grateful to the thousands of companies, individuals, and foundations for all the various ways they have supported the project.

However, even though construction is winding down, our fundraising campaign is still actively raising funds to ensure we have the resources to meet the goals of our campaign. Please visit our website or read our next newsletter for important updates.

Another significant change implemented in 2023 was making our doctor and nurse services more accessible to families by no longer charging for medical appointments. Instead, families are invited, but not obligated, to donate to support our mission. This change ensures that families are never turned away from our vital care based on their financial situation.

As we enter our 35th year, we reaffirm our collective commitment to the mission of the Clinic. This mission is not possible without the continued support of many people, including you.

Adam D. Heaps, MS, MBA
Executive Director

Participating in a Gene Therapy Trial

Noah's Story

"I've dealt with it my entire life," says Noah Crosby as he discusses his day-to-day life with a rare genetic disorder called phenylketonuria (PKU). PKU is a rare metabolic disorder that can cause a toxic buildup of an amino acid called phenylalanine in the brain, significantly affecting mental functioning and quality of life. Patients with PKU need to follow a special diet very low in protein, significantly limiting what they can eat throughout their lifetime. Newer medications allow some people with PKU to eat more protein, but they don't work for everyone. For Noah and other people facing PKU, however, there's a glimmer of hope on the horizon.

"As a boy when attending birthday parties, I would often need to bring my own cupcake. This is just one example of how PKU affected my childhood. When I reached adulthood, I spent years searching for solutions and reached a point where I was tired of dealing with it every day. I continuously researched new and groundbreaking PKU therapies and saw a gene therapy trial recruiting patients. I entered my information on the website and the Clinic for Special Children reached out to me. The Clinic was a clinical site for this gene therapy trial," says Noah.

The call from the Clinic was just the first of many steps to see if Noah would be a suitable candidate to participate in the study. "During the screening process, there were multiple tests I needed to clear to ensure that I would be able to participate. However, I kept on hoping that everything would be okay and that I would get the gene therapy someday," Noah says.

Thankfully, Noah met all of the screening requirements to become a participant. The long-awaited day of his gene therapy dosing came in June 2023. "The process of being administered gene therapy is surprisingly boring," exclaims Noah. Through a simple IV infusion, while sitting in a hospital bed at Penn Medicine Lancaster General Hospital, Noah was dosed with PKU gene therapy with Clinic physicians and researchers by his side.

Noah's decision to become a research participant was not hard for him to make. "My hope for joining the trial is to enjoy an ice cream cone with my son one day. I want to contribute to the field of science and make a better future possible for children with PKU. Growing up with PKU, I realized there are other kids out there who have a harder experience living with the disorder, and I want to help create better futures for them."

"I feel like I've experienced much more joy in my day-to-day life since receiving gene therapy," says Noah. "I have hope that one day PKU will be 'solved'. And for new parents having a child with PKU - understand that the future is looking bright. You can live a relatively normal life and hopefully, soon, a child receiving a diagnosis of PKU will have a completely normal life to look forward to."

Thank you to Noah for sharing his story and helping to create brighter futures for families facing rare genetic disorders!

PATIENT CARE *STATS*

1,738 *active patients*

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

4,530 *laboratory tests to help identify & manage genetic illnesses*

5,022 *patient encounters*


Hill-Rom
ADVANTA 2

Translational Research at the Clinic

Interview with Karlla W. Brigatti, MS, CGC | Research Director

A central tenet of the Clinic's mission is to translate insights and knowledge into actionable ways to improve the health of the families we serve. Karlla W. Brigatti, MS, CGC, Research Director, explains how research at the Clinic has evolved, why we practice research, and how we integrate science and medicine to provide better care.

Why is research important to the Clinic?

Research has always been central to the Clinic for Special Children. There is immeasurable value in understanding the conditions that present themselves through the patients and families who seek our care. As clinicians, researchers, and scientists, it's our responsibility to find answers to health-related questions that can change the quality of life for families. The heart of the Clinic's mission is to find unique solutions that directly improve patient care. We collectively see quite a few conditions that are sometimes very rare outside the Plain communities; those rare disorders do not attract the same attention as a common condition might. That does not make them any less significant for those affected families. I've heard a wise saying, "It's not rare if it's in the chair!" At the Clinic, we believe we are uniquely poised to address important needs for families through research that no one else can do. At its essence, this is research.

How has research evolved at the Clinic?

The Clinic's first projects revolved around the conditions that affected the families who came to the Clinic for care: GA1 in the Amish and MSUD in Mennonites. Careful study and lots of hard work enabled the clinicians managing these patients to improve treatment options like innovative formula design and protocols during hospital stays, dramatically changing the lives of those families. As our understanding of the genetic contribution to health and wellness has grown, the Clinic's research broadened to finding the genetic causes for the conditions we see. Now through tests like the Plain Insight Panel™ (PIP) we can identify genetic findings that impact health and are working hard to use that knowledge to find treatments that target the genetic cause (what we call "proactive") rather than the symptoms that prompt a visit to the doctor (known as "reactive" or symptomatic treatment).

As a genetic counselor, I have always been interested in the relationship between genetics and health. I joined the Clinic's team in 2014 to help with some of our research efforts, and I have never had a dull day! Over that time, we've been more systematic and intentional about how we collect data, communicate our findings, and follow the dynamic landscape of regulatory oversight. Over the past decade, the number of studies, complexity, and number of families affected by our research has increased substantially. The visibility of the Clinic as a leading expert in certain rare genetic conditions has also increased due to the number of our published publications. Recognizing this, the Clinic formally carved out the research department in 2018 and I was appointed Research Operations Director. Despite that growth, we remain grounded in the reason we do research at the Clinic - to answer important healthcare questions that directly impact the lives of the families we serve.

How are research projects chosen and prioritized?

Typically, a pressing clinical need prompts a research question for us to consider. For example, we are currently following patients with immunodeficiencies and are studying how we can best advise families on deciding one course of action for treatment versus another.

We look at how we can study the questions that present themselves to us in a systematic way that addresses them in a rigorous, ethical, and scientifically sound fashion. If we feel that we are uniquely positioned to answer a research question that can result in a better quality of life for our patients, we will prioritize it on our research agenda.

Why is the Clinic uniquely poised to conduct research?

We often see certain rare genetic conditions at a higher rate in the communities that we serve than you would find elsewhere around the world. An example is a disorder called TNNT1 myopathy, commonly known as Amish nemaline myopathy or "chicken breast disease." It is estimated to be found in about 1 in 1,000,000 in the general population while 1 in 400 babies born in the Lancaster Amish community have this condition. Because of our unique experience with this condition, it's incumbent on us to find answers that enable our patients

to have a better quality of life as a result.

As we think about the landscape of new therapies, we are always mindful of advocating the special circumstances of the populations that we serve. In other words, on the basis of Plain families being uninsured or uninsurable, we advocate for their access through patient access programs with pharmaceutical companies so there's more equity. We recognize the contribution Plain families make to the research community, and don't want them to be left behind with therapies they helped bring to market.

How are the Clinic's research findings applicable beyond the Plain community?

In the gene therapy space, we've been involved in several clinical trials recently. The disorders studied in these trials, spinal muscular atrophy (SMA) and phenylketonuria (PKU), are found relatively frequently in the Plain community but are also really common (for genetic disorders) around the globe. Our work in both of those disorders has a direct impact on patients living with those conditions worldwide. Our participation in these trials also advances the field of gene therapy as a whole.

What is a project that the Clinic is currently working on?

A specific project on the horizon is one led by Dr. Laura Poskitt that studies best practices for a disorder called congenital adrenal hyperplasia (CAH). Dr. Poskitt has taken a very thoughtful and focused approach on best practices for therapies for this condition. She's already shared her findings with other large healthcare systems, who are interested in utilizing her findings to create better practices for their own patients with this type of CAH.

What is a future research project that you're looking forward to?

We're excited to be part of a phase III study for a sponsored therapy for Fragile X syndrome. Fragile X is the most common genetic cause of intellectual disability worldwide. We see it at an increased rate in the Mennonite community. This condition has had multiple failed clinical trials to date, often because the studies were not well-designed. This new study we are participating in has promise to demonstrate durable therapeutic benefits in response to medication. With this study, we finally have the potential for a real impact on the patients we serve in the Fragile X community.

RESEARCH *STATS*

24 NEW *disease-causing genetic variants identified*

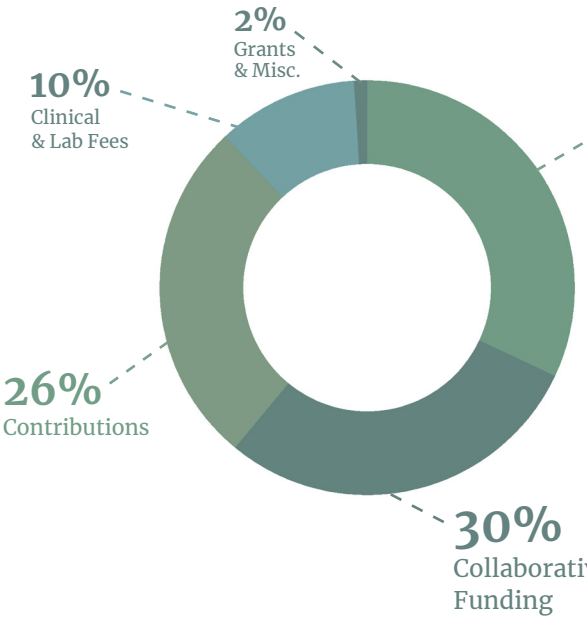
4 *peer-reviewed publications*

Financials Overview

A fiscal year in review

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 providing compassionate and cutting-edge care to families who need it most. Patient and lab fees made up only 10% of our 2023 operating budget, demonstrating how we're able to keep direct care costs affordable. The remaining 90% of the funds came from philanthropic sources like benefit auctions, fundraisers, contributions, and grants, as well as collaborative relationships. We truly could not do our work without the support of many people, like you! Thank you for allowing us to serve special children and families.

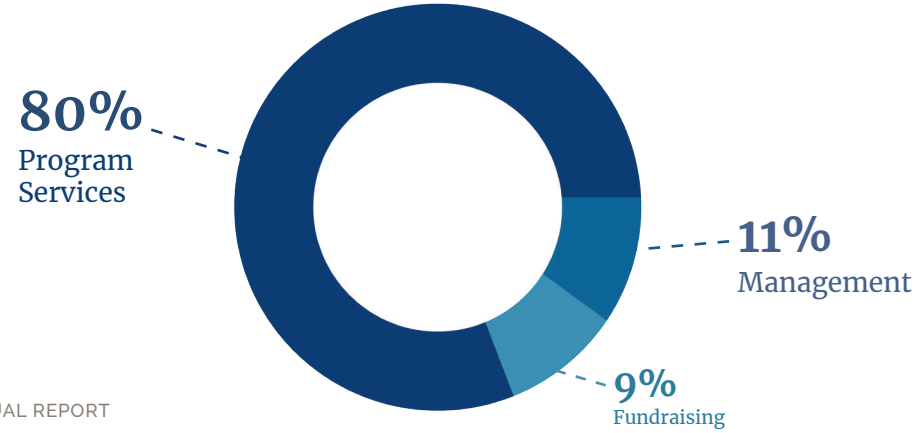
2023 REVENUE SNAPSHOT



60%

of CSC's revenue came from philanthropic sources like benefit auctions, fundraisers, contributions, and grants.

2023 EXPENSES SNAPSHOT



Donor Spotlights

Why do you support the Clinic?

Mr. John Thackrah

"I give to the Clinic because for many years I have seen the positive results of their efforts to help children born with life-threatening conditions. I live at Willow Valley Retirement Communities. They held a Holiday Bazaar and a group of residents sold note cards and pictures with all the proceeds going to the Clinic. This was a great opportunity to make people aware of the Clinic's work with children and tell them about the Clinic's new facility that will open in April of 2024. It was fun and a huge success."

John, pictured third from left, hosting a Clinic table at the annual Willow Valley Communities Holiday Bazaar.
Photo credit: Willow Valley Communities

High Foundation

"With a heart of service and a spirit of collaboration, High Foundation builds bridges to opportunity for individuals, organizations, and communities, striving for an equitable world of beauty, prosperity, and peace. We seek to be catalytic in our approach to funding, working to achieve the best possible outcomes for the organizations we partner with and support. In the case of the Clinic for Special Children, their new home in Leacock Township translates research into effective affordable medical care for the Plain community and beyond, a purpose that aligns nicely with the mission of the High Foundation."

FINANCIAL *STATS*

10%
of revenue came from patient and lab fees

$6.2 million
2023 operating budget

Keeping THE Promise

A Campaign for the Clinic for Special Children

What a difference a year makes!

Many important projects were in progress at the Clinic in 2023, and the largest was the construction of our new facility in Gordonville, PA! After years of working on identifying a site, land planning, township approvals, and more, our entire team was happy to see the project come to life this year.

At the start of 2023, excavating work was well underway and the footprint of the new Clinic was visible in the earth. By the end of 2023, we were in the final stages of construction on our new home (now planned to open in just a few short weeks in April 2024). A visual timeline below shows how the project's construction has progressed over the last year!

On Tuesday, April 4, 2023, the Clinic's 34th anniversary, we commemorated the start of construction on the Clinic's future home and formally announced our fundraising campaign with a groundbreaking and tree planting ceremony. A maple tree was planted in honor and memory of all the special families the Clinic has served over three decades and the continued work that will happen in our new facility. Idario Santos, the father of a son who is cared for by the Clinic, shared how much the new building will mean to the care of his family. Members of our staff, board, and capital campaign committee shared remarks on the importance


Capital campaign committee at the tree planting ceremony


April 2023


June 2023


early July 2023

of the day. It was exciting to finally share the news about the Clinic's future with many of our friends and supporters.

Over the summer months, the facility took shape right before our eyes! Volunteer construction crews erected steel beams, built walls, poured foundations, laid stone, installed decking, and more. Thankfully the weather cooperated and we kept our construction on track (and a bit ahead of schedule!) throughout the summer. By the end of August, the exterior construction of the building was almost complete and the installations of siding, roofing, and stonework entered final phases. Our staff, board, and capital campaign committee also got their first glimpses of the new building in August during behind-the-scenes tours, and the excitement built for our move in April 2024!

In September, the interior construction kicked off in earnest. The interior phases of construction progressed floor-by-floor and included HVAC, plumbing, electrical, drywall installation, insulation, painting, trim installation, cabinetry, and more. In December, the top floor was almost complete and the main floor and basement entered their final weeks of construction. We were very grateful that the project stayed on track throughout 2023 and progressed smoothly through each construction phase.

Capital campaign committee & board tour the new building

The support provided to our new building project this past year has left us incredibly inspired. Many supporters, including families, businesses, and community members have given their time, talents, and resources to secure the future of the Clinic. Gifts of in-kind materials and labor alone has represented over $1.7 million towards the fundraising of the new building.

While we still have some final funds to raise, we are hopeful we will finish our campaign strong this year. The support of many people in 2023 will allow us to keep our promises - to care for children as they age, seek the promise of life-saving therapies, and be here for families whenever they need our care.

late July 2023

November 2023

December 2023

Clinic *for* Special Children®

Our Staff

Amy Albright, MS, CGC
Genetic Counselor

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

Karlla W. Brigatti, MS, CGC
Research Director

Vincent Carson, MD
Pediatric Neurologist

Joelle Williamson Clark, MPH
Clinical Research Manager

Kelly Cullen
Marketing & Communications Manager

Skye Gawn
Gift Processing & Data Entry Coordinator

Jennifer Giacoio
Medical Receptionist

Adam D. Heaps, MS, MBA
Executive Director

Christine Hendrickson, RNC
Nurse

Candace Kendig, RMA
Office Manager

Lavina King
Community Liaison

Alanna Koehler, PhD
Assistant Laboratory Director

KaLynn Loeven
Laboratory Scientist II

Julia Martin
Development Associate

Alexis McVey, BSN, RN, CPN
Nursing Director

Grace L. Meier, MD
Family Medicine Physician

Laura Poskitt, DO
Medical Director

Erik G. Puffenberger, PhD
Laboratory Director

Stephen D. Ratcliffe, MD, MSPH
Senior Consulting Physician

Donna L. Robinson, MSN, CRNP
Nurse Practitioner

Ashlin Rodrigues, MS
Clinical Research Analyst

Sean Schreckengast
Laboratory Scientist I

Angela M. Sepela, MBA
Development Director

Dawn Sheets
Medical Receptionist

Jessica Snyder, PHR, SHRM-CP
Human Resources Generalist

Kevin A. Strauss, MD
Head of Therapeutic Development

Erin Sweigert
Research Associate

Anne Thomas, LPN
Licensed Practical Nurse

Sarah Thomas, RMA
Medical Assistant

William Van Ess, MS, CFE
Accounting Manager

Susan Walther, MS, CGC
Genetic Counselor

Kelly Woodson
Event Manager

Board of Directors

Jan L. Bergen

Cindy Bo, MBA
Secretary

Herman Bontrager
Chair of the Board

Elam Esh

Leon Hoover
Vice Chair

Leonard Hurst

Mark Martin
Treasurer
Chair- Finance Committee

Jacob Petersheim

Jonathan H. Salvin, MD

Stephen Tifft, MD

Glen Zimmerman
Chair- Development Committee

P.O. Box 500
Intercourse, PA 17534

tel (717) 687-9407
fax (717) 687-9237

ClinicforSpecialChildren.org