NEWSLETTER

Clinic *for* Special Children

CELEBRATING 25 YEARS

VOLUME I NUMBER 35 * LANCASTER COUNTY, PENNSYLVANIA * SPRING 2014

For our 25th Year we are reflecting on experience while also looking forward. As we share in this issue of the NEWSLETTER: We recently outlined a new Strategic Plan to give us direction. Renovations to our building improved useful space. A new garden created a place to think, soothe, and inspire for staff, patients and families. We happily welcomed Dr. Katie Williams and several others to our staff and sadly bid farewell to others. With growth and time there is change and new challenges walk through our door every day. We thoughtfully and creatively seek solutions to difficult medical problems, but we are always reminded of our purpose:

> "Special children are not just interesting medical problems, subjects of grants and research. Nor should they be called burdens to their families and communities. They are children who need our help, and if we allow them to, they will teach us compassion. They are children who need our help, and if we allow them to, they will teach us love. If we come to know these children as we should, they will make us better scientists, better physicians, and thoughtful people."
> D. Holmes Morton, Co-founder

1989

Patient Case Load: Approximately 100
Different genetic disorders treated in children: 12
Disorders diagnosed by the Clinic's lab: 4
Infants tested through Clinic's newborn screening for GAI: 400
Budget for Clinic's first year: $163,000 Staff: 3
Capital Investment for Equipment and Building: $554,000

2014

Patient Case Load: 1,008 Active; 2,608 Total
Different genetic conditions treated in children: 150+
Mutations diagnosed at the Clinic by molecular methods: 148
All Infants screened for GAI through state wide screening in US
Genetic mutations originally identified by the Clinic: 65
Budget for 2014: $2,854,000 Staff: 15

"PROMISES TO KEEP "

From articles written for the first Newsletter in 1990:

In 1988 Dr. Holmes Morton first recognized that Glutaric Aciduria (GA1) was a common cause of cerebral palsy among Amish children in Lancaster County. The concept of the Clinic grew as he obtained evidence that brain damage from the disease was preventable, if infants could be diagnosed before complications developed.

First attempts to find support for research on GA1 and start the Clinic were frustrating. In the Spring of 1989 the Morton's laid the ground work and formed a voluntary board of directors with Dr. Richard Kelley, filed incorporation paperwork and met with local families and community leaders. The first meeting with the Mennonite community was hosted by Amos and Nora Hoover at their Lancaster County farm on July 29, 1989. The Morton's were advised that a clinic seemed like a good idea and if they could get it started, support would likely come from the Mennonite community.

The problem was HOW to get it started? Dr. Morton's grant application to NIH for GA1 research was not funded. In retrospect this disappointment was a blessing as it lead to other possibilities with more lasting promise for the future Clinic. As the Morton's prepared to sign for loans, an article by Frank Allen in the Wall Street Journal brought significant interest and support from readers around the country.With the donations received from WSJ readers, Hewlett Packard's gift of GC Mass Spec diagnostic equipment and loan of temporary office space by Lancaster General Health at Willow Lakes Health Center, the Clinic for Special Children officially opened in January, 1990.

A NEW MODEL OF HEALTH CARE

Most of Dr. Morton's efforts in 1989-90 were focused on identifying children who had glutaric aciduria and establishing the first newborn screening program anywhere in the world to diagnose Amish infants with GA1 before the onset of symptoms of brain injury. Among the patients he saw the first year were also Mennonite children diagnosed with maple syrup urine disease (MSUD) who previously relied on major medical centers in Philadelphia for their care, with distance and cost as major barriers to care. Essential lab equipment in 1989 included a GC/mass spectrometer, donated by Hewlett Packard and an HP amino acid analyzer used to monitor maple syrup urine disease which was purchased with funds raised by several Mennonite churches and an anonymous donor. Children with other genetic conditions and their families began to find their way to the small Clinic. As the need grew, land was found, the building was raised, staff gradually added over the years and the work continues to unfold and expand.

Today Dr. Morton, Dr. Kevin Strauss Dr. Katie Williams and a talented, dedicated staff provide medical care for 1,008 children with complex medical problems related to 150 different genetic conditions that affect every organ system and range from highly treatable to invariably lethal. The Clinic was the first in the US to screen infants for GA 1 which led to state wide screening and eventually screening in all 50 states. With early diagnosis came improved care and healthy outcomes for many children. The development of the Clinic happened to parallel the progress of the Human Genome Project which opened vast potential for using more advanced methods for diagnostic options led by Dr. Erik Puffenberger and his team in the CSC lab.

Our work continues to evolve and to find solutions to caring for children with complex medical problems here and for others in faraway places who by necessity have discovered our clinic.

2014 Benefit Auctions

June 7- Union County, PA

June 27- Shippensburg, PA

July 12- Shiloh, OH

September 13- Blair County, PA

September 20- Lancaster County, PA

PLEASE JOIN US

June 7, Union County, PA

Families in **Union County, PA** will sponsor the third benefit auction for the Clinic on June 7th at the Buffalo Valley Produce Auction at 22 Violet Road in Mifflinburg. Quilts, sporting goods, furniture, sewing equipment, wood crafts, low protein pizzas and many other delicious treats will be featured at the auction. We are very grateful to the Hoover, Martin, Lapp, Horning, Weaver, Zimmerman, Nolt, Stoltzfus familes and many others for their time and effort to organize this auction to benefit the Clinic for Special Children. This support is very much appreciated.

June 27, Shippensburg, PA

The annual benefit auction for the Clinic in **Shippensburg, PA** will be held on June 27th. Thousands of donuts made that morning are usually sold out by early afternoon. Handmade toys, quilts, furniture, landscaping plants, a new carriage and a wide choice of food will be available for buyers. The Shippensburg Benefit Auction is now held every year on the **4th Saturday of June** instead of the third Saturday of July. Thank you to the Oberholtzers, Zimmermans, Leinbachs, and many other families in the Shippensburg region for this wonderful support.

July 12, Shiloh, Ohio

The Shiloh auction supports the Clinic for Special Children and the DDC in Middlefield, OH. Handmade quilts, furniture, crafts, farm supplies, garden items, and of course delicious food will be found at the Blooming Grove Produce Auction. Thank you to the Newswangers, Sauders, Burkholders, Martins and many other families who help with this auction.

September 13, Blair County, PA

Morrison's Cove Produce, Rt. # 36 South of Roaring Spring, PA

September brings two benefit auctions for the Clinic for Special Children. The first is in **Blair County, PA, September 13th** at Morrison's Cove Produce Auction on Route #36, south of Roaring Spring. In addition to quilts, furniture, crafts, chicken barbecue and other delicious food items, this sale always has a beautiful array of mums to get us ready for fall

September 20, Lancaster County, PA

Leola Produce Auction , Brethren Church Road, Leola, PA

The last and final auction of the year is in **Lancaster County on September 20th** at the Leola Produce Auction, BrethrenChurch Road, Leola, PA. Breakfast is available starting at 7:00 am and the auction opens at Block #1 at 8:30 am. Remarks by Dr. Morton, Dr. Strauss and Dr. Williams are scheduled at 11:30am with special items and quilt and furniture sales immediately following remarks. There will also be a silent auction block for many items including gift certificates for farm, home and midwifery services.

Among the special items to be auctioned this year are a special maple tree sculpture and individual maple leaves that are beautiful works of art hand forged by a local Amish owned iron works company (see article about the "Memory Tree"). Quilts include the Postage Stamp, Wedding Ring, Log Cabin, Trip Around the World and many other patterns. Carriages, farm supplies, outdoor furniture, swing sets, sheds, plants and many other items will be sold on four auction blocks.

THE MEMORY TREE

A Special Tree is Growing in White Horse.....

A unique maple tree sculpture is being forged as a special item for the Lancaster auction this year by a local iron works company located in White Horse. The **Memory Tree** sculpture is inspired by the Japanese Maple tree planted in front of the Clinic building when it opened in 1991. The tree was planted by the Morton's in memory of children with glutaric aciduria, maple syrup disease and other treatable genetic disorders who had died before the Clinic was established. The Japanese maple was chosen because it was the favorite tree of one of the mothers of children who are remembered.

With the Clinic's 25th Anniversary The **Memory Tree Project** seeks to honor the vision of Holmes and Caroline Morton and their fervent belief that children with rare, inherited disorders deserve local, accessible, state of the art medical care. This fundamental belief continues to guide the Clinic's mission and has led to major scientific advances for children everywhere with these disorders. The Clinic has grown to exemplify preventative medicine and the extraordinary economic benefits for the communities we serve.

The maple tree sculpture will be featured for bidding at the Clinic's Lancaster Benefit Auction on September 20th. The 3/4 scale sculpture is a unique work of amazing artistry. It would look spectacular in a large public foyer like that of a hospital or business or on the grounds of a college.

A duplicate of the tree will be created and donated to the Clinic for the new garden area.

MAKING THE MOST OF OUR SPACE

Over the past year the Clinic building was renovated to make better use of our space. As the staff and necessary equipment have expanded, we needed to make more room without any further additions to the existing building. Large rooms were divided to create more office space, the third floor conference area was redesigned to work for larger group meetings, old floors were refinished, more efficient storage for supplies was built. We are very grateful to a generous couple who donated funds to build a shed in our parking lot for visiting horses. The shed will provide shelter for at least two horses with buggies.

A NEW GARDEN FOR THE CLINIC

In late April a group of volunteers gathered to create a new garden space for the Clinic and also spruce up landscaping around the building. The design for the garden was contributed by a professional landscape designer and several professional landscapers (pictured below) were on hand to guide volunteers

in installing the new garden and plantings. All of the materials, plants, mulch and some of the supplies were donated. A new sitting wall to frame the garden was built by a local stone mason from stones gathered from a patient's father's field.

The second forged **Memory Tree** that is being created will stand in the center of the garden and will honor all special children - past, present and future. The garden area will be used as a place to meet, reflect or simply to enjoy the beautiful landscape that surrounds the Clinic.

OUR STAFF

WE WELCOME DR. KATIE WILLIAMS ~

After a two year search for 'just the right doctor', the Clinic was delighted to welcome **Dr. Katie Beth Williams** to our staff last November. Dr. Williams completed her pediatric residency at the University of Wisconsin in Madison where she also received her MD and previously, her PhD in Biochemical and Molecular Nutrition. She first learned about the Clinic when she attended a lecture given by Dr. Morton in 2012 at the medical school in Madison. Dr. Williams grew up among dairy farms in Wisconsin and together with her husband, Matt, and two young sons is very much at home among dairy farms in Lancaster County. We are very happy to welcome her as a vital member of the Clinic's staff.

We are very grateful for the assistance and generous support provided by **Lancaster General Health** for our search to find another physician and help with interview and relocation expenses

photo by Dan Marschka

In her own words, Dr. Williams describes her decision to join the Clinic's staff.

I heard many stories about the Clinic for Special Children during my residency program in Wisconsin. The physicians who cared for children with genetic and metabolic disorders at my training hospital would often talk about the work being done at the Clinic and look to the Clinic's research to help guide their patient's care. Just by chance, Dr. Morton happened to visit Wisconsin in October of 2012. I was fortunate to have the opportunity to hear his presentation to the medical staff and also attended a meeting with the Plain Communities in the area. It was quite powerful to hear about the impact the Clinic has had on the patients and families it serves. It was also quite evident that this work was deeply meaningful to families as well as the staff at the Clinic. It was a type of medicine I had not seen before and I was immediately intrigued. When Dr. Morton mentioned that the Clinic was searching for another physician, I only hoped I would be lucky enough to be considered for the position. Somehow, just over one year later, my family and I moved to

Strasburg so that I could be part of the Clinic. We are very grateful for this opportunity and have felt very welcomed by the staff and the families at the Clinic. I have much to learn but already find each day to be both challenging and rewarding; just what I had hoped for.

Katie Beth Williams, MD

WELCOME NEW STAFF:

Millie Young, R.N. joined the Clinic's staff as a pediatric nurse last September. Previously, Millie worked on the Pediatric service at Lancaster General Hospital and many of the Clinic's patients and families had the privilege of her attention as their nurse while at LGH. We are very pleased to welcome Millie as part of our staff and many patients and families are happy to see a familiar person at the clinic.

Yalonda Kosek recently assumed her job as the new medical receptionist. If you have called our office since April 1st., you probably spoke with Yalonda. She brings her experience as a medical office receptionist, scheduler, front office organizer and problem solver to our office along with her dedication and commitment to the clinic's mission.

STAFF FAREWELL:

Rebecca Smoker retired in December from her role as part-time Office Manager. She was the first person hired by the Morton's when the Clinic opened for patients in January, 1990 at the temporary Willow Lakes location. Rebecca's deep understanding of the local Amish and Mennonite communities and her familiarity with many family networks were essential to the Clinic's early success in reaching families and children who needed our services. Rebecca's help in managing the office, answering phones, scheduling patients, ordering supplies, interpreting information to families and wise counsel were invaluable to all of us, especially Holmes and Caroline Morton. She is missed and her unique gifts will never be replaced. We are very grateful for her dedication and commitment to the Clinic for the past 24 years.

STAFF PROGRESS:

Erica Eisenbise is now Office Manager, filling the roles previously served by Rebecca Smoker and Miriam Echternach. She previously was the Office Coordinator and Receptionist.

Adam Heaps was appointed part time Administrative Director on April 1st. He assumed these duties in addition to his position in the lab as Laboratory Scientist.

Caroline Morton, the Clinic's co-founder, has gradually shifted her responsibilities as Executive Director to other staff members (administration and development) and a contracted accounting firm (financial and payroll). She will continue to work at the Clinic part time on special projects and on the strategic direction of the Clinic.

Dr. Holmes Morton will resume work at the Clinic in June refreshed after a year's sabbatical that gave him time to work on special clinic projects and time to recover from some surgery.

A CLINIC GROWS IN BELLEVILLE

One of the projects Dr. Morton has been working on is to help establish a new clinic in the central region of Pennsylvania that will serve the special needs of children and adults. The Central Pennsylvania Clinic (CPAC) will operate as a separate organization from CSC with its own funding, staff and volunteer board of directors. Dr. Morton's brother, Paul, has been involved in helping to lay the groundwork for the new clinic. Support for the project has been growing and volunteers organized the first very successful benefit auction in May to help raise funds for the project that will complete the matching challenge from the University of Pittsburgh Medical Center (UPMC). The CPAC board hopes to hire a core staff in the next few months to begin to provide services at an existing site in Belleville until plans for a new building can be developed.

A colorful parking lot at the Belleville CPAC Auction

Genomic Medicine & the Plain Populations of North America Conference #2

The second conference on Genomic Medicine & the Plain Populations of North America will be held on August 15 and 16 in Cleveland, OH, coordinated by the Das Deutsch Center in Middlefield, Ohio. The Clinic for Special Children sponsored the first conference last July to review diagnosis and management of genetic diseases in Plain populations in North America. The conference will address topics such as:

- An overview of genetic disorders prevalent in Plain populations in various regions, of the U.S. and Canada,
- Review of newborn screening approaches and compare follow-up programs,
- Discussion of population specific methods available for genetic testing,
- Information on current research projects
- Progress of new clinics.

OUTREACH & RESEARCH

CSC continues to schedule outreach clinic days to see patients in **Somerset County** every three months and in **Belleville** once a month.

CSC has recently focused on three topics of clinical research designed to improve scientific understanding and clinical management of genetic disorders. The three disorders are **Congenital Adrenal Hyperplasia (CAH), GM3 Synthase Deficiency** and **Propionic Acidemia (PA)**. Updates on CAH studies are presented in **Abby Benkert**'s article in this issue. Research on GM3 is reported by **Joshua Wesalo** in his article. CSC hosted meetings for families of children with these disorders to update them on recent progress. Studies on Propionic Acidemia will conclude this summer with recommendations for improved management of the complex disorder.

In Partnership with F&M: CAH HANDBOOK

In collaboration with the Clinic students and faculty from **Franklin & Marshall College** developed ***Congenital Adrenal Hyperplasia: A Parents' Handbook.*** The handbook serves as an informative guide designed to help families with children with congenital adrenal hyperplasia (CAH) understand their child's condition. The handbook explains CAH in a simplified, accurate and approachable manner, with an awareness of the biological complexity of CAH and careful consideration of parental care. It includes information about CAH support groups. The Howard Hughes Medical Institute and F & M College provided support for the project.

2014 Recent Events:

•FEBRUARY:

Dr. Strauss was a Visiting Professor at Jefferson Medical School.

•MARCH:

Dr. Morton presented a poster on Propionic Acidemia treatment at the meeting of the Society for Inherited Metabolic Diseases in California.

Dr. Strauss lectured about the Clinic's healthcare model at the meeting of the Alliance of Independent Academic Medical Centers in San Diego.

•APRIL:

GM3 Family Day provided an opportunity for families of children with GM3 to meet with physicians and researchers to hear updates on efforts to find a successful treatment for this debilitating disorder.

CME Seminar for Midwives: Routine Care for the High Risk Neonate featuring presentations and case studies by Dr. Morton, Dr. Strauss, Dr. Williams and Dr. Puffenberger from the CSC and Dr. Mitchell Kresch, Hershey Medical Center, Dr. Kevin Lorah and Dr. Philip Bayliss, Lancaster General Health. Approximately 40 midwives attended from Pennsylvania.

Dr. Strauss lectured at the Genetic Metabolic Dietitians International Conference in Dallas, TX on the acute and chronic management of Maple Syrup Disease.

•MAY:

TEDx Lecture presented in Lancaster by **Dr. Strauss**.

Dr. Strauss traveled to Brazil to speak with families and medical representatives who are engaged in establishing a clinic inspired by the model of the Clinic for Special Children.

PKD Family Meeting was held with **Dr. Morton** in the Belleville area to review information regarding upcoming preliminary trials for a potential new treatment for patients with Pyruvate Kinase Deficiency. It is hoped the new medication will provide more effective management of PKD to avoid the difficult complications of the disease. The study will be based at CPAC in Belleville.

Coming Up:

July 8: **Transplant Review Day** cosponsored with Pittsburgh Children's Hospital Liver Transplant Department.

July 18: Dr. Weiner Orthopedic Consultation Day

August 8: Yoder Dystonia Family Day

August 15 and 16: Genetic Medicine in Plain Populations Conference, Cleveland, OH

September 18: CNTNAP2 (also referred to as CASPER 2) Family Day.

WE NEED YOUR SUPPORT !

We want to make every effort to keep services at the Clinic affordable for all families, especially those who do not have insurance. The cost of running the Clinic and providing care has grown significantly in the last few years. Our fees are still significantly lower than charged elsewhere for comparable services. Amino acid analysis at CSC is $75 while the charge at a nearby medical center is $750. Community sponsored benefit auctions generally raise about one-third of the funds needed to meet the actual cost of providing medical services. The balance needed usually comes from donations from individuals, grants and foundations.

We use a small portion of the Research & Education Endowment Fund each year to help fund research and educational programs. Our long term goal is to increase the Research & Education Endowment Fund through major gifts to cover a more significant percentage of our research, education and clinical studies costs.

Major gifts will help us grow the Research & Education Endowment Fund to support the Clinic's work. Please consider a gift to help us expand and sustain our work. With our THANKS.

34% Charitable Gifts

13% Investment Income

22% Fees for Services

31% Auction Proceeds

2013-2014 RESEARCH FELLOWS

During the past year the Clinic was privileged to have two Research Fellows working on projects that have a direct impact on the care of patients. **Abby Benkert,** the current Avery Fellow, is completing her studies on Congenital Adrenal Hyperplasia and plans to apply for medical school. **Joshua Wesalo** has divided his time with the Clinic and Franklin & Marshall College with his research on GM3 Synthase Deficiency. He will attend the University of Pittsburgh Medical School as a MD/PhD candidate.They each describe their work and what the year has meant to them.

Joshua Wesalo, Research Fellow, F&M '13

Many individuals in the Old Order Amish population carry a genetic mutation that can cause GM3 Synthase Deficiency in their children. Although affected children are normal at birth, they suffer from intractable seizures, deafness, and poor brain development in infancy. As children and teenagers, these individuals lose control of their movements, suffer from profound mental retardation, and become blind. These individuals typically die before adulthood.

Because of their genetic mutation, children suffering from GM3 Synthase Deficiency cannot produce a chemical called GM3. Every cell in your body contains GM3. Chemically, GM3 comprises a type of sugar and a lipid joined together. The lipid portion of GM3 anchors GM3 into the outer membrane of your cells, and the sugar hangs off like a flag that tells nearby cells what to do. The "sugar flag's" signaling function plays an essential role in orchestrating brain development and maturation, which explains the devastating neurological symptoms in these children.

Dr. Strauss has hypothesized that treating these children with seven to ten grams of GM3 per year could allow them to live normal lives. We tried supplementing the patients' diets with a buttermilk concentrate that contains appreciable amounts of GM3 (0.56% by weight) earlier this year, but the children did not absorbing enough GM3 to make much of a difference.

Thanks to support from the Clinic and from Franklin & Marshall College, my alma mater, I have had the opportunity to try to help these families by spending the year developing a procedure for preparing an injectable form of pure GM3 using organic chemistry. The process involves building up the sugar and lipid portions of the molecule (the flag and the flagpole) separately, and coupling them together in one final step. So far, we have made tremendous strides in the process. We validated our coupling procedure using simpler versions of the GM3 sugar and GM3 lipid. At present, we are only a few steps away from preparing actual GM3 at a scale suitable for treating several infants.

I have also spent several weeks at the Clinic working to revive a scientific instrument called an UHPLC (ultra-high-pressure liquid chromatograph) that Scientific Systems, Inc./Lab Alliance donated to us. This instrument takes solution containing a mixture of chemicals, separates the components, and measures each one. We use less-advanced system, an HPLC, to measure blood levels of amino acids for many of our patients quickly and economically. The UHPLC has the potential to speed up the amino acid measurements, which take about 30 minutes, to 3 minutes. I'm also looking into using the instrument to measure blood levels of fat-soluble vitamins (vitamins A, D, E, and K), and to size up fragments of DNA to help with our genetic testing. This work has been a refreshing change-of-pace. Organic synthesis work resembles very painstaking cooking, but for work on the UHPLC, I get to break out the torque wrench and become a mechanic troubleshooting a maze of pumps, valves, and tubing.

I'm extremely grateful to the Clinic for having had the opportunity to work here this year. Since last May, I've been on a journey applying to MD/PhD programs. In evaluating applicants, these programs value deep experiences at the intersection of science and medicine. A lot of students pursue such research at big-name academic medical centers or at the National Institutes of Health, but I can't think of a better place to do this than Strasburg, PA.

Many investigators in the biomedical sciences spend their entire careers studying a disease without ever meeting a patient. My work at the Clinic gave me the opportunity to meet GM3 families early-on in my research. Getting to know several parents and meeting their children has motivated me immensely. Every time another chemical reaction fails and I have to redo it or rethink my approach, I think of them and think of my promise that I'd work my hardest to give them hope. This fuels me through long nights in the lab, and I hope I'm lucky enough to have the opportunity to serve patients with science and medicine again in the future.

Abby Benkert, Avery Fellow 2013-2014

My eyes never really adjust to the darkness of early morning as I walk into patients' homes carrying my "blood draw box." By the time I leave the house, after drawing blood from children affected with congenital adrenal hyperplasia (CAH), the sun is rising over the barn and the rooster signals the day's arrival. I listen to the hum of the portable centrifuge in the backseat, at this intersection of technology and rural farm life.

During my year as the Avery Fellow, I have been studying CAH and conducting a clinical study trial comparing two different treatment options to determine whether giving one dose of dexamethasone before bedtime will improve overall health and disease control as compared to standard hydrocortisone therapy. While the children are on each treatment, I measure several key compounds, including the steroids that can accumulate as a result of CAH, and compare the results of each treatment method. Over the past seven months, I have completed the study and analyzed results for a few patients, and am now currently collecting data on the remaining CAH children.

In addition to this clinical research project, I am developing laboratory methods so the Clinic can analyze the blood samples "in-house" to monitor CAH. Currently, patients must come to the Clinic to have their blood drawn, and those blood samples are then sent to another laboratory; which means these tests are expensive and receiving the results can take up to a few weeks. Like the way in which the Clinic monitors MSUD, our hope is that CAH can be monitored from filter papers that families can mail to the Clinic.

I'm excited for when this project will be nah faddich – all done – so I can share my results with the families and recommend the safest and most effective treatment for their children's CAH, as well as eventually provide a cheaper and faster way to monitor the disorder.

Besides learning about clinical care and scientific research (of which I've learned a tremendous amount!), I have come to understand the meaning of community. The idea of community permeates every corner of the Clinic. The community of people who helped build the Clinic in a day, who come to the annual benefit auctions to support and celebrate the Clinic's work; the community of clinicians and laboratory scientists working together to solve complex problems; the community of CAH families who I have come to know and appreciate during my clinical research study; the community of all who walk through the doors of the Clinic. And at the heart of this community are God's special children – who teach us about disease, who teach us to be selfless and work together towards improving health, who, most importantly, teach us how to be a community. I have been so blessed to have been a part of the Clinic community during my time as the Avery Fellow and to have worked with all the wonderful CAH patients and families. I know that I will take my experiences and all that I've learned from this special medical home with me on my path to becoming a physician and into my own medical practice

LOOKING AHEAD

STRATEGIC PLAN 2014-2020

I. OUR MISSION

Provide comprehensive local medical care, integrate the science and the practice of medicine, and share knowledge to improve the health of children who suffer from genetic disorders.

II. OUR VISION

We envision the Clinic for Special Children as a medical home for Amish and Mennonite (Plain) children 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 children have access to the most timely, affordable, and effective health care. 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 in the future.

III. PRECEPTS

- Accurate and cost-effective diagnosis.
- Comprehensive, accessible, and affordable care.
- Deep understanding of disease mechanisms.
- Integration of basic research and clinical practice.
- Preemptive diagnosis and disease prevention.
- Strategic collaborations to enhance patient care and research.
- Education and community empowerment.

IV. STRATEGIC GOALS

GOAL 1. Strengthen the medical home.

A *medical home* is a system of care that is informed, comprehensive, coordinated, compassionate, and culturally effective. The CSC will provide a medical home for Amish and Mennonite children who live within reasonable commuting distance and suffer from genetic conditions within the CSC's areas of expertise. Medical home services include routine pediatric care, core diagnostic and analytical laboratory services, design of specialized diets and treatment protocols, emergency and inpatient care, coordination with specialists, community education, and public health initiatives.

Objectives:

- Provide outpatient clinical services pertinent to the needs of the core patient population.
- Develop capacity for additional disease-specific analytical testing on-site.
- Systematize screening for genetic variants of medical relevance.
- Expand on-site subspecialty services as determined by need.
- Develop capacity for 4-person call rotation.
- Collaborate with LGH Pediatric Hospitalist Service to coordinate inpatient care.
- Solicit feedback about service quality from families who use CSC as a medical home.

GOAL 2. Develop an effective consultation service.

The CSC will provide *consultation services* for patients who live far away, are not children, or have conditions outside the staff's core expertise. Consultation services include affordable diagnostic evaluations, coordination of care with regional providers, and referral to appropriate specialists.

Objectives:

- Create triage process to improve utilization of CSC resources.
- Standardize diagnostic process for new patient evaluations (fee structure, turnaround, reporting).
- Develop network of specialty providers and institutions.
- Develop network of collaborating out-of-state primary care providers.

GOAL 3. Collaborate to improve patient care.

The CSC will seek out and maintain *patient-centered* partnerships focused on clinical care, basic research, and public health.

Objectives

- Expand clinical capacity through collaborations: e.g. additional specialty services on-site, tissue transplants, etc.
- Expand research capacity through collaborations: e.g. affordable molecular testing, complex data analysis, pathophysiology research, public health research, epidemiology.
- Seek collaborations focused on population genomics and public health.
- Hire/appoint a *Clinical and Research Coordinator* to manage key projects and relationships.

GOAL 4. Educate families, clinicians, and students.

The CSC will engage in a broad range of educational activities designed to *build capacity* for better pediatric health services both within and beyond CSC's primary catchment area. Educational activities should: 1) enable parents to better care for their children, 2) empower communities to create local medical services, and 3) train young clinicians and scientists to serve future generations.

Objectives

- Establish protected professional time for education and outreach activities.
- Educate parents through an active program of Family Education Days and parent-focused publications.
- Promote community awareness of actionable genetic health risks.
- Enable medical home services in other communities through education.
- Assist in education and training of LGH Family Practice residents. • Provide *basic science* and *public health* research opportunities for F&M faculty and students.

GOAL 5. Fund clinical, research, and education missions.

The CSC will develop *fundraising goals* that allow the institution to 1) diversify funding sources; 2) grow the operating budget to support additional staff, 3) build new sources of funding for clinical and laboratory research; and 4) fund educational activities.

Objectives

- Link fundraising to *specific* clinical and research priorities
- Grow the combination of clinic fees and annual gifts by ≥ 25%
- Develop a program of planned giving.
- Target major gifts to sustainable endowments (total endowment goal $10M).
- Actively seek private foundation gifts.
- Judiciously seek grant opportunities that benefit CSC patients.
- Seek funding opportunities through collaborations.

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CELEBRATING 25 YEARS

Clinic *for* Special Children

MISSION

Provide comprehensive local medical care, integrate the science and the practice of medicine, and share knowledge to improve the health of children who suffer from genetic disorders.

CLINIC FOR SPECIAL CHILDREN
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The Clinic for Special Children is a non-profit 501(c)(3) tax exempt organization and a registered charitable organization in Pennsylvania.
Tax ID# 23-2555373

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Caroline S. Morton, EdM., Founding Director
Kevin A. Strauss, M.D., Medical Director
Katie E. Williams, M.D., PhD. Pediatrician
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