



Clinic for
Special Children

Newsletter

ISSUE #47

SUMMER 2019



Searching cross-country for a cure The Newkirk Family

Kris and Maureen Newkirk experienced an easy pregnancy and delivery with their third son, Charlie, in December of 2015. They never expected that several days after Charlie was born his state newborn screen test would come back positive for a disorder they had never heard of - Glutaric Acidemia Type 1 (GA-1). The Newkirk family soon learned that only four other people had been diagnosed with GA-1 in their home state of Arizona. GA-1 is a genetic disorder in which the body is unable to break down essential amino acids, which can lead to irreversible brain damage.

At four-days-old, Charlie started treatment at Phoenix Children's Hospital with a strict diet of a special formula just for GA-1 patients in order to prevent brain damage. Over the next year and a half, Kris and Maureen conducted a great amount of research on GA-1 and quickly realized limited information was available. During their research efforts they kept finding the name of Dr. Kevin A. Strauss and the Clinic for Special Children.

The Newkirk family decided to take a leap of faith and travel to Pennsylvania to meet with Dr. Strauss to learn more about GA-1. Their first appointment at the Clinic included

a three hour meeting with Dr. Strauss where he explained the Clinic's research and many years of experience with GA-1. Maureen and Kris said of that first meeting, "We walked away with the feeling that Charlie was going to be okay. There's something magical about coming down the lane to the Clinic with its reassuring feeling - it's so different than anything out there."

The Newkirk family says that each year gets easier as Charlie is allowed more protein and stress the importance of the newborn screen test. Unlike when Charlie was younger, they are able to treat him at home when he is sick, and don't have to visit the hospital. Today, Charlie is a happy three-year-old and enjoys playing with his older brothers, who are always looking out for their little brother.

In addition to being a patient family, Kris and Maureen Newkirk have been amazing supporters of CSC along with the Crain Family Foundation (check out our Donor Spotlight section in this issue to read more about their efforts!) and are advocates for giving back to the rare disease community. We look forward to seeing the Newkirk family each summer when they visit the Clinic!



SPECIAL DATES

Ohio Benefit Auction Saturday, July 13

Blooming Grove Produce Auction
Shiloh, OH
7 a.m. - 4 p.m.

Missouri Benefit Auction Saturday, August 17

Ed's Machinery
Memphis, MO
9:30 a.m. - 4 p.m.

Labor Day - Office Closed Monday, September 2

All Day

Blair County Benefit Auction Saturday, September 14

Morrison's Cove Produce
Roaring Spring, PA
7 a.m. - 4 p.m.

Clinic for Special Children 5k Saturday, September 21

The Clinic, 9 a.m. - 11 a.m.
Application and Details Inside

Community CSC Benefit Dinner Wednesday, September 25

Martindale Fellowship Center
4 p.m. - 7 p.m.
Details Inside

2019 Extraordinary Give Friday, November 22

Online Fundraiser - All Day

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

Staff News

Yalonda Kosek

Join us in congratulating Yalonda Kosek, Office Coordinator, who graduated in May with a degree in computer information systems and information technology. Yalonda joined CSC in 2014 and was promoted to Office Coordinator in 2017. She is responsible for coordinating front office activity, patient billing, and serving as a consultant for some of CSC's IT operations. Congrats, Yalonda!



Caitlin Russell

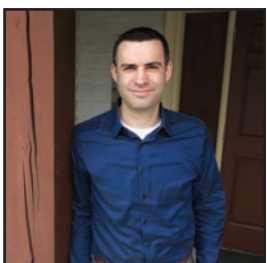
Caitlin is joining CSC in July as a Genetic Counselor. In this role, Caitlin will assist in new patient triage and intake, providing genetic counseling services, and managing the logistics of the molecular diagnostics service.



Caitlin received her Master's degree in Genetic Counseling and Master's in Public Health from the University of Pittsburgh. In the summer of 2018, Caitlin worked alongside Karlla Brigatti, MS, LCGC at CSC as a summer intern. Caitlin explains why she's passionate about the Clinic, "CSC's commitment to serving as a liaison between innovative genetic research and families so that they may wisely make important health decisions epitomizes why I became a Genetic Counselor."

William Van Ess

Bill joined CSC in April as an Accountant. In this role, Bill will manage invoicing, bank reconciliations, maintaining the general ledger, and all facets of ac-



counting for the organization.

Bill earned his Bachelor's Degree in Accounting from Albright College in Reading, PA and his Master's Degree in Accounting from Neumann University in Aston, PA. He spent several years as a Certified Fraud Examiner. Bill appreciates the work of CSC in helping children, as he has his own young daughter.

Save the Date!

2nd Annual Clinic for Special Children 5k Run/Jog/Walk

**Saturday, September 21
9:00 a.m. – 11:00 a.m.**

at the Clinic for Special Children

Calling all runners, joggers, and walkers! Join us for our 2nd annual Clinic for Special Children 5k on Saturday, September 21st, presented by Nemours Children's Health System! Last year our first race had over 280 runners and raised over \$24,000! Registration is now open for the race- an application is included with this newsletter and is also available at the Clinic's website (www.ClinicforSpecialChildren.org/events/5k). This year's event will feature a kid's fun run (new!), silent auction, bake sale, children's activities, and more!

Register by 9/2 to receive a discount!

Mail the application in this newsletter with payment to CSC (Attn: Kelly Cullen) or visit ClinicforSpecialChildren.org/events/5k to register online. If you would like additional applications or flyers, to volunteer during the event, or to sponsor the 5k, please contact Kelly Cullen by calling 717-687-9407.

CSC Community Benefit Dinner

**Wednesday, September 25
4:00 p.m. – 7:00 p.m.**

at the Martindale Fellowship Center

Join us for a community benefit dinner at the Martindale Fellowship Center (352 Martindale Rd., Ephrata, PA 17522) supporting the Clinic for Special Children! The cost for the event will be on a donation basis. The menu will feature oysters, shrimp, ham, cole slaw, rolls, green beans, fruit cups, and soft ice cream. Through-

out the dinner, there will be talks by CSC staff and patient families on topics such as new treatments, carrier testing, and more!

To RSVP for this event, please contact the Clinic at RSVP@clinicforspecialchildren.org or call 717-687-9407.

Vaccination Clinics Baby Shots Available

We offer a vaccination clinic every 3rd Tuesday of the month from 9:30 a.m.-4:00 p.m. at CSC. To participate, your child does not need to be a patient of the Clinic but you must not carry health insurance. The clinics are \$20 per child per visit (no matter the number of vaccinations) and are open to children aged 18 and under.

If you would like to schedule your child for a vaccination clinic, please call the us at 717-687-9407 to set up an appointment.

A confirmed case of measles has been reported in Lancaster County. Measles is preventable with a vaccine. If you or your children have not received the measles vaccine, you need to get vaccinated. If you choose not to vaccinate yourself or your child, understand the risks and responsibilities.

Looking for Sponsors! 2019 Extraordinary Give

**Friday, November 22
Midnight-11:59 p.m.**
ExtraGive.org

We're already gearing up for the 2019 Extraordinary Give, Lancaster's largest day of online giving! Last year the Clinic raised over \$65,000 during the ExtraGive's 24-hour giving period!

We are looking for companies that would be interested in sponsoring "giving matches" for our 2019 ExtraGive. This would allow anyone who gives to CSC during the ExtraGive to have their donated dollars matched and will increase giving potential. Your company's name and logo will be featured on CSC's giving page and promoted leading up to the ExtraGive (if desired). If you are interested in this opportunity, please contact Kelly Cullen at the Clinic at 717-687-9407 or kcullen@clinicforspecialchildren.org.



Dr. Laura Poskitt

A new physician joins the CSC team!

We are excited to announce that Laura Poskitt, DO, will be joining our clinical team in July! Dr. Poskitt, a trained Pediatrician, committed to joining the Clinic for Special Children team in 2018 during her last year of residency at Nemours/A.I. duPont Children's Hospital in Wilmington, DE. She is a native of Lancaster County, having graduated from Hempfield High School. She attended Grove City College for her undergraduate work, and graduated medical school from the Philadelphia College of Osteopathic Medicine.

"I know that being open to learn makes you the best teacher, so I'm also hoping to learn from patients and families as well."

When asked about joining CSC, Dr. Poskitt said, "I think the thing I'm most looking forward to is being back where I grew up and having the opportunity to invest in those most vulnerable children in Lancaster County and those that come to CSC to receive care. One thing that I love about medicine is the opportunity to develop trusting relationships where we help each other- hopefully I can provide some help to children and I know in return I will learn and grow so much. The CSC offers so much through working both with the staff and the exceptional patients and families we care for."

During this past year, Dr. Poskitt was a Chief Pediatric Resident at Nemours/A.I. duPont Hospital, which she explains has taught her to be a more effective teacher. Her training as a DO (Doctor of Osteopathy) also makes her passionate about the healing of the whole person, not only focusing on the concrete medical needs, but also exploring the social, mental, and emotional aspects of health as well.

Dr. Poskitt and her husband, Will, have a son Oliver who is 1-year-old. In her free time, she enjoys reading historical fiction, traveling to new places, and visiting art museums. We're excited to welcome Dr. Poskitt to the CSC team!

Giving Opportunities

TWO Local Foundation Giving Matches!

Each dollar given will be matched by a local foundation

General Support

We are excited to announce our first giving match of 2019, a **1-for-1 match up to \$15,000**. The match is available thanks to the continued generosity of a local family foundation and is for wherever the funds can be best used. This match opportunity expires on December 3, 2019!

Electronic Medical Records Upgrade

The second giving match opportunity is a **1-for-1 match up to \$25,000** provided by The Steinman Foundation to upgrade our outdated electronic medical records (EMR) system to Epic - a cutting-edge, universal, and secure system offered through partnership with Lancaster General Health.

This EMR update is integral to the Clinic's ability to offer best possible health outcomes by allowing patients to access their records, communicate effectively with other healthcare systems, and maintaining a secure network.

To direct your gift to one of the matches, select the appropriate box on the included giving envelope or call CSC at 717-687-9407.

Mark your Calendars for the remaining 2019 Benefit Auctions!

7:00 a.m. Breakfast | 8:30 a.m. Auctions Begin
Physician Remarks and Quilts to Follow

The annual CSC benefit auctions and charitable donations make up approximately 40% of the Clinic's annual revenue! This support makes it possible for the Clinic to continue to provide affordable, specialized care to children and adults living with rare genetic disorders not just in Lancaster County, but across the country and the world!

Ohio Auction | 7.13.19

Blooming Grove Auction Inc.
1091 Free Road
Shiloh, OH 44878

Contact: Leon Newswanger | 419-896-3336

Missouri Auction | 8.17.19

Ed's Machinery
17920 US Hwy 136
Memphis, MO 63555

Contact: Harlan Burkholder | 660-341-4113

Blair County Auction | 9.14.19

Morrison's Cove Produce
174 Windy Acres Lane
Roaring Spring, PA 16673

Contact: Mervin Z. Martin | 814-793-3530



“There is no footprint too small to leave an imprint on this world.”

Cherished Lives | Palliative Care Program

By: Keturah Beiler, RN

My name is Keturah Beiler and I work at the Clinic for Special Children as a Registered Nurse. I am also currently in school to complete my Bachelor of Science in Nursing (BSN). I have been working at the Clinic for a little over 2 years. My first year I worked part-time and since January 2018, I've been working full-time at CSC. In addition to providing nursing services alongside Dr. Kevin A. Strauss, Medical Director at CSC, it is my privilege and honor to provide home visits for babies that have been born with a lethal genetic diagnosis.

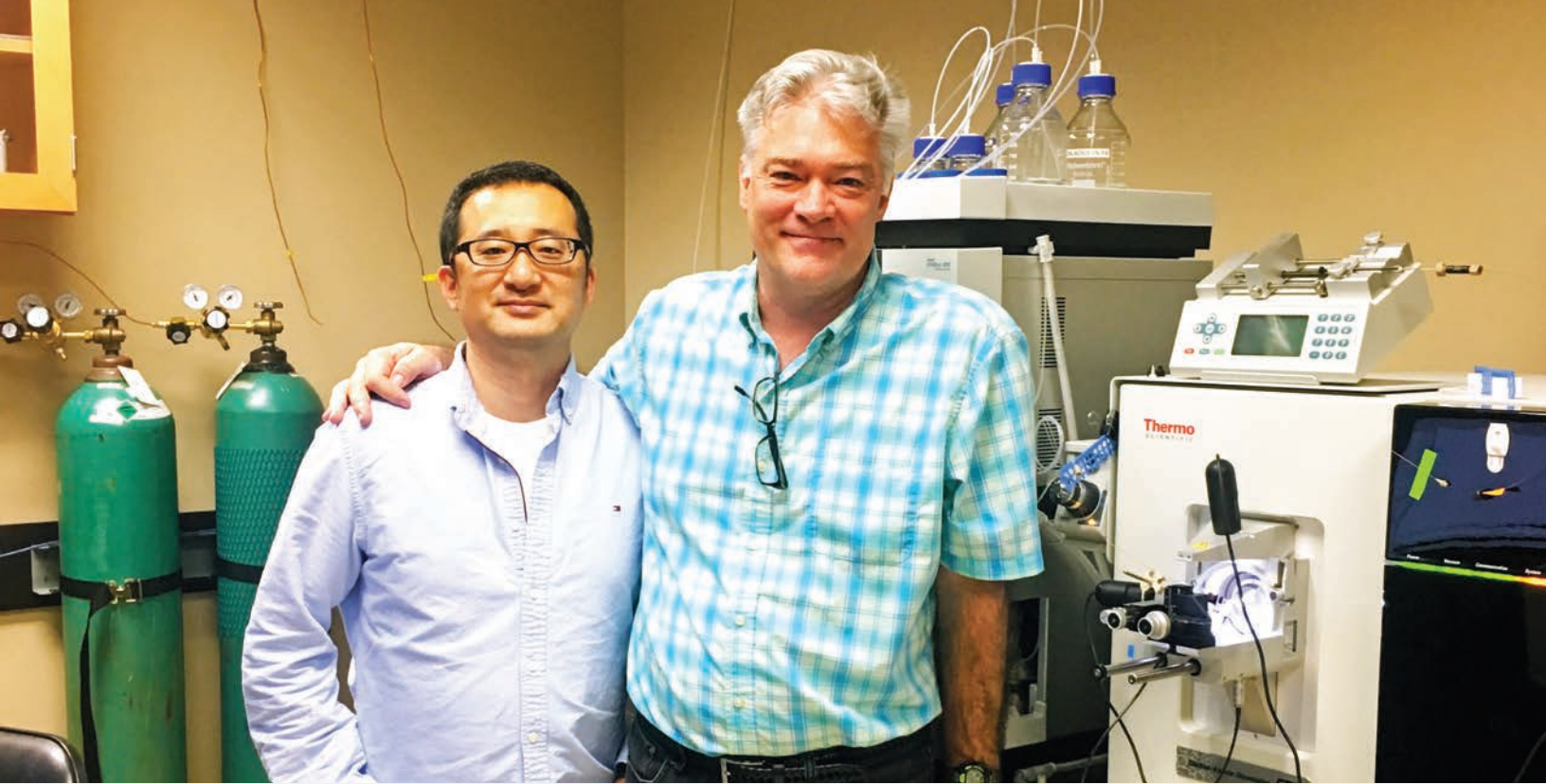
At CSC, it is our desire to provide supportive care for these babies and to walk alongside the family by providing support during a life-changing and difficult time. As a nurse, I am drawn to work with families as they prepare to part with their little one. When a family reaches out to CSC about a baby with a condition that indicates a lethal diagnosis, like Amish Lethal Microcephaly, our team will often see them in their home to establish a care plan. Most often this plan arranges for them to be seen in their home once a month or as needs arise.

The Clinic is also committed to providing support and comfort to families who have children with severe, life-limiting diseases. These diseases are those in which we know the diagnosis but do not have a cure, and the disorder will most likely shorten their life span. Families choosing comfort care, primarily at home, is part of a model of care known as palliative care. This type of care makes patients as comfortable as possible without using extreme interventions in the hospital.

Although CSC has informally provided palliative care for many years, we recently formalized the program. We now call it “Cherished Lives”. This name reflects our view that every life, even a short one, should be cherished. We all have much to learn from every patient we treat.

As part of the Cherished Lives program at CSC, we have begun to make memory gifts for the families that lose little ones to rare genetic disease. At one of our first visits, we stamp the baby's feet onto paper to record their footprints. After their passing, we make small quilts with their footprints printed on the fabric as a token of remembrance and comfort for the families. Log Cabin Quilt Shop & Fabrics in Bird-in-Hand, PA has graciously donated supplies for the project. Priscilla Beiler, the mother of Lavina King, Community Liaison at CSC, pieces the wall hangings and quilts to make a finished product. Since the start of the quilt project, families have expressed their gratitude for this gift to remember their cherished baby.

The main objective of our Cherished Lives program is to clearly learn what parents' goals are for their children related to their care. This is a discussion that families can have with the doctors here at CSC. Our long-term goal for our new Cherished Lives program is to expand the services for these families to include home visits by a nurse, care packages, and other ways to reduce the suffering of these children and their families. I'd like to thank Dr. Vincent Carson, Pediatric Neurologist at CSC, for his leadership with this program.



Collaborator Spotlight: University of Georgia | Complex Carbohydrate Research Center

Pictured above (L to R): Kazuhiro Aoki, PhD and Michael Tiemeyer, PhD

When Dr. Michael Tiemeyer was completing his undergraduate work at Johns Hopkins University, he was exposed to an emerging field that would come to be known as Glycobiology (the study of carbohydrate function and structure). His PhD graduate work at Johns Hopkins School of Medicine provided him with “a deep appreciation of glycolipid biochemistry and glycolipid function in neural tissue.” After obtaining his PhD, Dr. Tiemeyer joined the Complex Carbohydrate Research Center (CCRC) at the University of Georgia and currently works as the Co-Director of the CCRC.

The CCRC is unique as it has the highest concentration of research scientists with expertise in understanding the function and structure of complex carbohydrates, a class of molecules that regulate almost every aspect of cell survival and function. Dr. Tiemeyer explains, “by being in a center filled with scientists that share common research goals and complementary expertise, we are able to make rapid progress in novel research areas.”

During Dr. Tiemeyer’s early years at CCRC, on the other side of the globe, Dr. Kazuhiro Aoki was obtaining his PhD degree in life science at Kyoto University in Japan. His thesis focused on studying the role and structure of glycoconjugates in pathogenic fungi, explaining the mechanisms of host-pathogen interaction via glycoconjugates. After completing his PhD, he joined Dr. Tiemeyer’s team at CCRC as it’s known as one of the best institutes in the world to study function and structure of complex glycans. Dr. Aoki currently works as a Senior Research Scientist at the CCRC.

The CCRC team was first introduced to the Clinic for Special Children (CSC) through Steven A. Roth, PhD, an entrepreneur in the biotechnology field and a faculty member at the University of Pennsylvania, when he visited the CCRC. At the time, the CCRC team was conducting research on a disorder called GM3 synthase deficiency, and Dr. Roth connected the CCRC with Dr. Kevin A. Strauss, Medical Director at CSC. It turned out that GM3 synthase deficiency was a disorder that had been researched at CSC for many years. This was the beginning of a long-standing and innovative collaborative rela-

tionship between the CCRC and CSC.

The CCRC, CSC, and the Plain Community Health Consortium (PCHC) recently published the largest clinical description of ST3GAL5 (GM3 synthase) deficiency. The study examined objective measures of biochemistry, auditory function, brain development, and caregiver burden of GM3 synthase deficiency. Throughout this study, the CCRC “provided analytic assistance by measuring blood levels of glycolipids in PCHC patients and unaffected individuals using advanced mass spectrometric methods that the CCRC developed and published with PCHC collaborators. We are also investigating the basic cell biology of neuronal survival using cultured cells that have the same biosynthetic defect as a patient with GM3 deficiency”, explains Dr. Tiemeyer.

The CCRC and CSC also recently published a paper describing an accurate method of measuring the amount of GM3 and other gangliosides in various samples. This method was used to generate data for the clinical description paper and can be used to measure the effectiveness of future treatments.

Dr. Aoki describes the CCRC & CSC’s collaboration, “I have been studying glycomic consequences of human disorders in cancer, rare genetic disorders and one of the projects targets on a rare mutation in ST3GAL5, GM3 synthase which causes ‘Salt and Pepper Syndrome’ (S&PS) in African American siblings. The S&PS is allelic to the Amish GM3 deficiency.” When approached about partnering with CSC and PCHC, Dr. Aoki said, “I am always wondering if my research contributions support people suffering from health problems. By taking the chance to work with CSC and PCHC, now I have obtained the great opportunity to interact with patients, doctors, clinicians, and other scientists through the project by providing my scientific expertise which may contribute in the development of new therapies for patients with GM3 deficiency.

Drs. Tiemeyer and Aoki have been able to attend the annual GM3 family days several times, and enjoy meeting everyone there. We thank Drs. Tiemeyer and Aoki for their innovative work and we look forward to continuing our partnership!



Our Staff

Keturah Beiler, RN
Nurse

Lauren Bowser
Research Fellow

Karlla Brigatti, MS, LCGC
Research Operations Director

Vincent Carson, MD
Pediatric Neurologist

Kelly Cullen
Communications Manager

Adam D. Heaps, MS, MBA
Executive Director

Christine Hendrickson, RNC
Nurse

Candace Kendig
Medical Receptionist

Lavina King
Community Liaison

Yalonda L. Kosek
Office Coordinator

KaLynn Loeven
Laboratory Scientist

Julia Martin
Development Assistant

Laura Poskitt, DO
Pediatrician

Erik G. Puffenberger, PhD
Laboratory Director

Donna L. Robinson, CRNP
Nurse Practitioner

Ashlin Rodrigues
Laboratory Scientist

Caitlin Russell, MS, MPH
Genetic Counselor

Emily Seitz
Scientific Grant Writer

Kevin A. Strauss, MD
Medical Director

William Van Ess, MS, CFE
Accountant

Millie Young, RNC
Research Nurse

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Donor Spotlight

The Crain Family Foundation

After Jesse Crain retired from playing Major League Baseball (MLB) for many years, he and his wife, Becky, knew they wanted to give back to the communities that supported them throughout their lives. With this initiative, Jesse and Becky created The Crain Family Foundation through the Players Philanthropy Fund, a 501(c)(3) public charity that enables athletes, entertainers and other philanthropists to collect tax-deductible contributions that support any charitable cause. In addition to giving back, they wanted to show their kids how to make a difference and leave a legacy in the world. At the onset, they knew they wanted to support charities that help children and families, focus on baseball initiatives, and generally help people in need. Since its inception, the Crain Family Foundation has supported organizations throughout Arizona, Minnesota, Colorado, Pennsylvania, and the entire country!

The story of how the Crain family became involved with the Clinic for Special Children started when they met the Newkirk family, as their kids were on the same little league baseball team coached by Jesse. Kris Newkirk, father of Charlie, began telling Jesse about Charlie's journey with GA-1 and CSC (see front page story) and asked if he could sell bracelets at the event to help raise money for families with rare disease. Soon after, Jesse invited Kris to their first 'Swing Fore the Kids' fundraiser event. Since that first event, the Crain Family Foundation has included the Clinic for Special Children as a participating organization! The 'Swing Fore the Kids' event is held annually in February close to Rare Disease Day (and Spring Training!) at TopGolf in Scottsdale, AZ and features celebrity appearances, a silent auction, great food, golfing, and more! Each year the event grows larger with this year's event raising over \$27,000 for the Clinic!

When asked why they are passionate about CSC, Jesse and Becky explained, "We are very grateful to have three children that are happy and healthy and we can provide beyond what they need and want. Since we have lived in different parts of the country, we have seen that not everyone is fortunate to have that. These families look okay on the surface, but once you dig deeper and hear what they go through, it hits home. We appreciate what we have and do what we can to help families and organizations to, at the very least, bring awareness."

In the future the Crain Family Foundation has a goal of getting the MLB involved in the various events and initiatives they put on each year. They're hoping to do something similar for Rare Disease in what various sports do for Breast Cancer Awareness (athletes wearing specific colors for awareness, promotions, etc.). They are currently looking for formal office space and hope to expand the connections they have throughout the country to help benefit the organizations close to their hearts. We thank Jesse and Becky for their amazing work on behalf of rare disease and CSC!



30TH ANNIVERSARY CELEBRATION

Thank you to everyone that joined us for our 30th Anniversary Celebration at the Clinic on Tuesday, April 16th!

*It was a fantastic day with great food, fun activities, historical mementos on display, and amazing company!
We hope you enjoy the photos below from the event.*





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“To serve children and adults who suffer from genetic and other complex medical disorders by providing comprehensive medical, laboratory, and consultive services, and by increasing and disseminating knowledge of science and medicine.”