



Clinic for
Special Children®

Newsletter

ISSUE #52

SPRING 2021



Finding Hope with TNNT1 myopathy The Stoltzfus Family

When Esther and Michael Stoltzfus were expecting their second son, Joseph (Josie), they knew it was a blessing to have a healthy baby as both of their families had a history of rare genetic disorders. When Josie arrived six weeks early in the spring of 2019, he spent his first 11 days in the NICU under observation because of his premature birth.

When he was born, he exhibited a strong cry and fed really well. However, shortly after Josie came home from the NICU, the Stoltzfus family noticed he started to have tremors. Josie's grandmother mentioned to Esther that tremors are commonly seen in babies with TNNT1 myopathy (also called 'Chicken/Pigeon Breast Disease' or TNNT1 for the affected gene). TNNT1 myopathy is a devastating rare genetic disorder that affects the muscles and their ability to contract and relax (read more on page 4).

When Josie was about eight weeks old, Esther and Michael noticed the tremors started to worsen and called the Clinic for Special Children (CSC) to make an appointment. "We wanted peace of mind as we felt that something might not be right", Esther explains. At their first appointment, they met with Dr. Vincent Carson, Clinical Operations Director at CSC, who explained that Josie's symptoms did resemble tremors commonly seen in babies with TNNT1 myopathy. A rapid diagnostic test was ordered and ran in CSC's in-house lab. The Stoltzfus family went home to await the results.

Later that afternoon, Dr. Carson called the Stoltzfus family to confirm that Josie's test result came back positive for TNNT1 myopathy. Right away the family and CSC developed a care plan for Josie and he was enrolled in our palliative care program, Cherished Lives. Keturah Beiler, Nurse & Cherished Lives Program Manager at CSC, visited the Stoltzfus family's home every other week to ensure Josie was comfortable and the family had what they needed.

Josie was also enrolled in our WiTNess research study (read more on page 5). "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." When asked about CSC's impact on Josie's life, Michael explains, "I don't know where we would have been without CSC. Anytime we needed something, you were there."

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. 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," the Stoltzfus family explains.

To us at CSC, Josie was a special boy with a bright personality that taught us so much as caregivers, researchers, and parents. The Stoltzfus family will always be a part of our CSC family, and we are grateful to them for sharing their story to help others.



SPECIAL DATES

Memorial Day - Office Closed Monday, May 31

All Day

Union County Benefit Auction Saturday, June 5

Buffalo Valley Produce Auction | Mifflinburg, PA

Lancaster County Benefit Auction Saturday, June 19

Leola Produce Auction | Lancaster, PA

Shippensburg Benefit Auction Saturday, June 26

Cumberland Valley Produce Auction | Shippensburg, PA

Ohio Benefit Auction Saturday, July 10

Blooming Grove Auction Inc. | Shiloh, OH

Missouri Benefit Auction Saturday, August 21

Ed Good Family Farm | Memphis, MO

Blair County Benefit Auction Saturday, September 11

Morrison's Cove Produce | Roaring Spring, PA

Clinic for Special Children 5k Saturday, September 18

At the Clinic & Virtual

Mark your calendars and save the date for our
4th annual 5k run/jog/walk!

As the situation around COVID-19 evolves, the event dates mentioned above could be subject to change.

Announcements from Clinic for Special Children®

Staff News

Keturah Beiler, RN



Keturah recently completed her studies at Eastern Mennonite University and earned her bachelor's of science degree in nursing!

Keturah joined the Clinic in 2017 and is an integral part of our clinical team. She works as the Cherished Lives palliative care program manager and as a Nurse at the Clinic.

Congratulations, Keturah!

Taylor Brown, RN, CPN



We're excited to welcome Taylor Brown, RN, CPN to the Clinic! She will work as a Nurse within our clinical team to provide exceptional patient care.

Taylor graduated from West Chester University with her bachelor's of science degree in nursing. In her free time, Taylor enjoys spending time with family, friends & her dog, hiking, and working out. A fun fact about Taylor - she is a self-proclaimed donut enthusiast!

Welcome to the Clinic, Taylor!

Jennifer Giacoio



We're excited to welcome Jennifer Giacoio to the Clinic as our Medical Receptionist! In her role, Jennifer will provide administrative support for the

Clinic's staff, patients, and visitors. She will help in triaging calls, scheduling patient appointments, updating records, and providing a welcoming environment at the Clinic.

She earned her bachelor's of science degree in Human Development and Family Studies from The Pennsylvania State University. In her free time, she enjoys painting, spending time with her family, and baking.

Welcome to the Clinic, Jennifer!

Donna L. Robinson, CRNP



In October of 2020 we celebrated a milestone work anniversary for our Nurse Practitioner, Donna Robinson! Donna celebrated her 25th anni-

versary at the Clinic! Donna has been an integral part of our clinic team for a quarter of a century and we feel grateful to work with her everyday.

At the Clinic, Donna is responsible for the clinical care of patients both in the office and hospital, taking call responsibility, interpreting amino acids, and coordinating patient care. She has helped with numerous Clinic research projects and contributed to many peer-reviewed papers.

Thank you for your dedication, Donna!

Important COVID-19 Updates

Vaccine

We have reviewed the available data from studies on the authorized COVID-19 vaccines, which show the vaccines are both effective and safe. The COVID-19 vaccine is now available for individuals who are 16 years old or older. Please contact your doctor with questions regarding the COVID-19 vaccine. If you are interested in being vaccinated, please call the Pennsylvania Department of Health at 877-724-3258 or visit their website for more information.

For individuals in or near Lancaster County, you can contact the WellSpan Health COVID-19 hotline at 855-851-3641 or for information on Lancaster's community vaccination clinic visit VaccinateLancaster.org or call 717-588-1020.

Office Operations

We continue to require mask wearing and temperature screening for all visitors to our clinic facility. If you have a scheduled appointment and are feeling unwell, we ask that you call us at 717-687-9407 before coming to your appointment. Our clinic staff is now back to operating at full capacity, as our staff are fully vaccinated for COVID-19. If you have any questions before your visit, please contact us at 717-687-9407.

Save the date!

4th Annual Clinic for Special Children 5k Run/Jog/Walk

Saturday, September 18th

9:00 a.m. - 11:00 a.m.

at the Clinic for Special Children

We are excited to announce our 4th annual Clinic for Special Children 5k! Strollers and wheelchairs are welcome! Registration will open in the summer - applications will be available in our next newsletter and on the Clinic for Special Children website.

More event details will be included in our next newsletter and the event is subject to change due to COVID-19 restrictions.

If you are interested in becoming a sponsor for the 2021 Clinic for Special Children 5k, please contact the CSC Development Team at 717-687-9407 or giving@clinicforspecialchildren.org.

We hope to see you at our 5k this year!

2021 Benefit Auctions Website

www.ClinicAuctions.org

To stay up-to-date on 2021 benefit auction happenings, visit the auctions website at www.ClinicAuctions.org! This website will be updated throughout the benefit auction season this year with any event changes, sale bills, flyers, and more.

2021 Mugs for Sale



Meet your new favorite mug! Our 2021 Clinic for Special Children mugs are now available for purchase! Each mug is just \$10 and available for purchase

on our online web shop (www.ClinicforSpecialChildren.org/store), in person at the Clinic, at 2021 CSC events, or by giving us a call at 717-687-9407.

This year's new tall mug features a unique charcoal and speckled design with a terra cotta bottom. It's perfect for your favorite coffee, tea, or hot cocoa!

ClinicforSpecialChildren.org

2021 Benefit Auction Season

7:00 a.m. | Breakfast

8:30 a.m. | Auctions Begin

Physician Remarks and Quilts to Follow

Union County Auction | June 5

Flower Sale and Rib Dinner | June 4 | 5-8 p.m.

Buffalo Valley Produce Auction

22 Violet Road

Mifflinburg, PA 17844

Contact: Leon Hoover | 570-966-2414

Lancaster County Auction | June 19

Leola Produce Auction

135 Brethren Church Road

Leola, PA 17540

Contact: Mark Martin | 717-733-3070

**Breakfast starts at 6:30 a.m.*

Shippensburg Auction | June 26

Cumberland Valley Produce Auction

101 Springfield Road

Shippensburg, PA 17257

Contact: Elvin Oberholtzer | 717-532-9088

Ohio Auction | July 10

Blooming Grove Auction Inc.

1091 Free Road

Shiloh, OH 44878

Contact: Michael Newswanger | 419-896-2184

Missouri Auction | August 21

Ed Good Family Farm

10507 County Road 813

Memphis, MO 63555

Contact: Harlan Burkholder | 660-341-4113

Blair County Auction | September 11

Morrison's Cove Produce

4826 Woodbury Pike

Roaring Spring, PA 16673

Contact: Mervin Martin | 814-793-3529



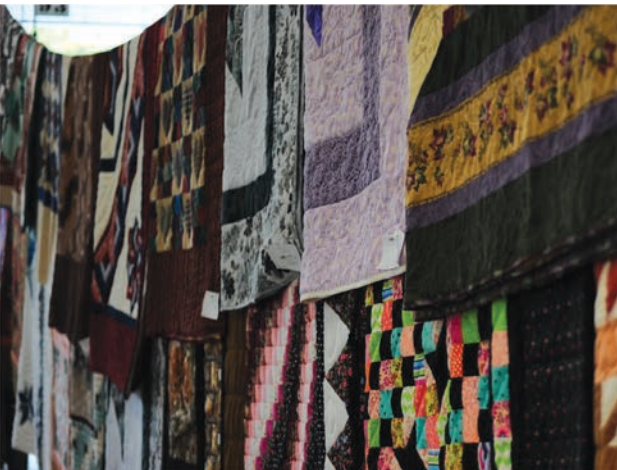
2021 Auction Season

Each 2021 auction will feature a day of handmade and unique items for sale, delicious food, and more!

The six benefit auctions make up about 26% of the Clinic's annual operating budget, which allows us to continue providing life-saving care to those with rare genetic disorders.

Please visit our auctions website (www.ClinicAuctions.org) to view the most current auction information.

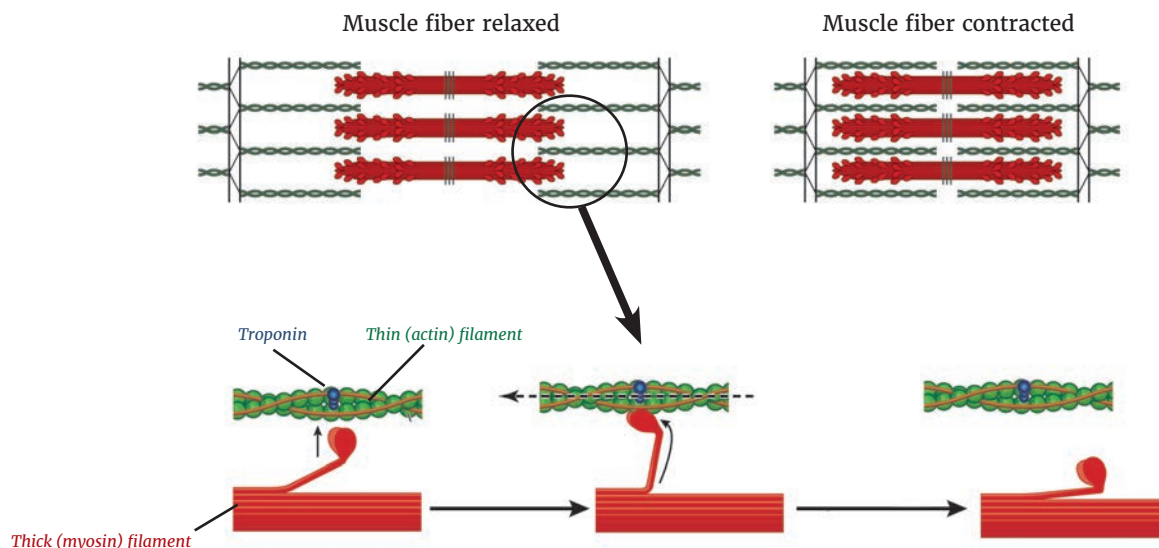
We recognize the rapidly evolving situation with COVID-19 and have been taking all precautions to keep our staff and patient families safe. Auctions are subject to change due to COVID-19 restrictions.



“The Clinic continues to grow in its mission to serve children and their families. And by working together, we have had the opportunity to touch many lives in many ways.”

– Kevin A. Strauss, MD | Medical Director

Excerpt from his remarks at the 2018 Lancaster County auction



The above illustration is a microscopic view of what happens when our muscles contract. Troponin (a group of proteins) allows the thick myosin filament in our muscles to attach to the thin (actin) filament when we have a muscle contraction. The actin and myosin interact with each other to mediate muscle contractions.

Troponin has a critical role in regulating the roles of actin and myosin and couples them to nerve signals to the muscles. In TNNT1 myopathy, this process is disrupted as described in the article below.

What is TNNT1 myopathy?

A devastating rare genetic disorder

TNNT1 myopathy (also called ‘Chicken/Pigeon Breast Disease’ or *TNNT1* for the affected gene) is a form of nemaline rod myopathy, a genetic disorder that affects about 1 in every 500 babies in the Amish community, but has been found in individual children all over the world. It causes progressive muscle weakness. It’s a devastating genetic disease and babies with this disorder usually die by two years of age from complications they develop as a result of the condition.

Signs & Symptoms

Several of the first signs and symptoms of TNNT1 myopathy include jaw and leg tremors and shakiness that start in the first weeks of life. The tremors and shakiness eventually go away. Over time, large muscles in the baby’s body gradually get weaker and their shoulder and hip joints become stiff. Around one to two years of age, the muscles that control breathing begin to weaken, which leads to difficulty breathing and more respiratory issues like pneumonia. They also develop the characteristic “chicken breast” shape to their chest. We can also see that this muscle weakness leads to scoliosis (curvature of the spine), which further affects breathing.

Disease Progression

TNNT1 myopathy is progressive, meaning that as the baby gets older, their symptoms will get worse. Infants will have trouble rolling over and sitting up because of their muscle weakness. Usually children are still able to grab and hold objects, are socially normal, and can learn like other children. Eventually, TNNT1 myopathy will lead to breathing trouble from a small chest and weak rib cage muscles.

Cause of TNNT1 myopathy

Muscles are made up of long, thin rods called myofibrils. Inside the myofibrils are small fibers that grab each other and then push using a group of proteins called troponin. When many of these fibers grab and

push all together, the whole muscle moves/squeezes.

In TNNT1 myopathy, muscle fibers grab onto each other, but then they get stuck and cannot keep moving because the troponin proteins cannot let go. Over time, this leads to muscle weakness and stiffness (reference image above for an illustration of this process).

How is TNNT1 myopathy diagnosed?

TNNT1 myopathy is caused by the lack of the troponin I protein in the muscle cell, due to an error in the *TNNT1* gene that directs the production (like a recipe) for the normal troponin I protein. Patients with TNNT1 myopathy have a genetic error in both copies of the *TNNT1* gene they receive from their parents (one from their mother, the second from their father). At the Clinic we have a genetic test that looks for the specific genetic error in the *TNNT1* gene and make a diagnosis when both copies of the gene have that change.

The Future of TNNT1 myopathy

We are currently conducting a natural history study for TNNT1 myopathy, called WiTNNESS, to learn more about the natural course of this disorder. This natural history study will allow us to measure the effectiveness of a therapy, if one becomes available (more about the WiTNNESS study on page 5).

We are also working with other medical and scientific experts in TNNT1 myopathy, so we have the best understanding of this condition from the way it affects patients to pinpointing how muscle cells change when they don’t have the troponin I needed for proper function. This involves a network of dedicated specialists across many countries, working in laboratories and clinics. We are working with scientific collaborators on developing a gene replacement therapy for TNNT1 myopathy, and the laboratory studies to demonstrate if and how it might work for the condition are happening right now.

WiTNness Natural History Study

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

Learn more about the WiTNness study below in an interview with our Research Operations Director, Karlla Brigatti. You can view page 4 of this newsletter to learn more about TNNT1 myopathy (also called ‘Chicken/Pigeon Breast Disease’ or *TNNT1* for the affected gene).

What is the WiTNness study?

WiTNness is an opportunity for us to learn more about the natural course of *TNNT1* myopathy in patients who have the condition as they are diagnosed, which usually occurs soon after birth. This study allows us to carefully measure the many different ways the condition affects patients over time, to capture the course of the disease as it happens in concrete ways rather than looking back and relying on memory. For this reason, we call this a “natural history” study, since it essentially allows us to “witness” the disease course in a precise way.

For example, if we want to know more about how breathing is affected for children with *TNNT1* myopathy, we can check the oxygen levels in their blood every few months, or measure the size of their chest rather than trying to remember in hindsight when breathing first became an issue. This gives us a more reliable and reproducible way to understand the disease course, and serves as the benchmark for evaluating the effectiveness of new therapies as they are developed. I like thinking of WiTNness as the timber frame to the house we are building where we can do something to dramatically change the course of the disease.

Who is being enrolled in the WiTNness study?

Our lab identifies between three and five babies with *TNNT1* myopathy every year. Usually this is because parents are known carriers of the condition and can have children with *TNNT1* myopathy, so they choose to do the genetic testing using umbilical cord blood of their newborns. In other cases, the baby has features of *TNNT1* myopathy, like tremors, or doesn’t seem to be as active or strong as other babies, so they are diagnosed with *TNNT1* myopathy after becoming symptomatic. Either way, we work with the families one-on-one to give their children the best care for the highest quality of life through our Cherished Lives palliative care program. We also talk to them about the WiTNness study and how their participation may help other *TNNT1* myopathy families in the future. We recognize that being a part of the WiTNness study may not change much for these children now, but we share the goal of getting to a therapy that will treat children in the future.

We are part of a network of medical providers and centers where other *TNNT1* myopathy patients have been identified and are treated. This includes other non-Amish families around the United States and in many other countries around the world. Those providers are also doing the same types of measures with their *TNNT1* myopathy patients that we capture in WiTNness, so we are able to see how *TNNT1* myopathy affects those children and their families as well. Not surprisingly, we are finding that they are very similar to our experience. We expect we will continue to add patients across the world to this group.

Why are you focusing on this disorder?

Until recently, therapies for rare disorders tended to focus on managing symptoms or working around the medical issue affecting growth and development, like giving a child with muscular dystrophy steroids to keep them able to work for longer periods of time. We also knew that this symptomatic approach in *TNNT1* myopathy has not worked well and didn’t improve the lives of these children with the condition. However, over the past few years, we have begun to see how gene-modifying therapies—those that correct the specific genetic problem that gives rise to the disorder—are a game-changer for some genetic conditions. We call these “gene therapies” for this reason, and we have seen them work in patients with spinal muscular atrophy (SMA) a disease with

many similar features to *TNNT1* myopathy, and for those patients to grow and thrive once they have been treated despite having SMA. We really believe that a gene therapy could work for *TNNT1* myopathy, and we are working hard with other *TNNT1* myopathy experts at other centers to develop this therapy.

Since babies with *TNNT1* myopathy are essentially normal at birth, we know we have a window of ideal time to treat these children with a therapy so we stop the disease from progressing. Gene therapy introduces a working copy of the *TNNT1* gene to the muscle cells where it can make the important *TNNT1* protein needed for proper muscle contraction and development. If we give the cells the genetic recipe to do this (though gene therapy), and do so early in life, we think we can protect these children from the serious features of *TNNT1* myopathy.

Why is this study important?

That’s a great question— if we are developing a therapy for *TNNT1* myopathy, why study it closely when no therapy is available? Well, WiTNness is getting us ready for a future clinical trial. Again, it’s the frame to a strong house we’ll have with a good therapy. It gives us the baseline we need, measures we can use and look to change when we give a therapy, and allows us to measure that therapeutic response in an informative and consistent way. For example, our current understanding with *TNNT1* myopathy is that these babies aren’t able to sit up on their own. If we introduce a therapy that allows these children to achieve that milestone, we would have a clear sign that the therapy is working.

What does the future look like?

WiTNness is a study that is part of a larger vision at the Clinic to find ways to help patients with genetic conditions overcome limitations in treatment options and provide them with better outcomes and quality of life. If we can develop a therapy for *TNNT1* myopathy patients, it is a victory that opens the door for a similar approach in other rare genetic conditions. Everyone wins! As we learn more about these therapies and they become more and more a part of the therapeutic landscape, I predict we’ll see this approach for many other disorders we see at the Clinic.

While there are no guarantees and we still have a lot of questions to answer and challenges to overcome, we are learning quickly as we move forward. We have high hopes that all this work will pay off in a therapy for *TNNT1* myopathy patients, but we don’t have a clear answer when that might be—except to say not soon enough! We are working with our partners and other experts to get to that finish line, but we have to really show that any therapy we develop will make a big difference, and that can take time. All I can say is that our team is as dedicated and determined as ever to help find solutions for our *TNNT1* myopathy families that gives their children with the condition the best quality of life that we can provide. It is humbling, inspiring, and at times really tough to take this journey with our families, and we are honored they’ve entrusted us to do so. We take that responsibility very seriously as we continue the good fight.



Karlla Brigatti, MS, CGC
Research Operations Director



Dr. Jin visiting Antarctica

Collaborator Spotlight

Jian-Ping Jin, M.D., Ph.D., Professor of Physiology and Biophysics, University Illinois at Chicago College of Medicine

When Clinic researchers and collaborators published the first research paper on TNNT1 myopathy in 2000 (also called ‘Chicken/Pigeon Breast Disease’ or *TNNT1* for the affected gene), Dr. Jian-Ping Jin contacted a co-author on the paper, Dr. Thomas Crawford. During their discussion, Dr. Crawford invited Dr. Jin to visit the Clinic in Strasburg to understand the condition and the community it affected. After the visit, Dr. Jin was inspired to start his important laboratory work focused on TNNT1 myopathy, solving the mystery of how changes to the *TNNT1* gene and the protein it encodes lead to the features of TNNT1 myopathy.

Dr. Jin is currently Professor of Physiology and Biophysics at The University of Illinois at Chicago College of Medicine. He is a graduate of Fourth Military Medical University, China, and University of Iowa. He completed his residency in cardiovascular medicine and postdoctoral research fellowships at University of Texas at Austin and University of Alberta, Canada. Most recently he was Professor and Chairman of Department of Physiology at Wayne State University School of Medicine in Detroit.

Over his long career in science, Dr. Jin has had one major purpose. As he put it, “The primary goal of my work is to enrich our scientific knowledge to improve human health.” A leading expert in the field of muscle contractility and cell motility (the way that a cell moves from one location to another in the body), Dr. Jin has published over 180 research papers during his career. He is the sole editor of two books: *Troponin: Regulator of Muscle Contraction* (Nova Science Publishers, Inc., 2013) and *Troponin: Informative Diagnostic Marker* (Nova Science Publishers, Inc., 2014).

His current research interest is in genetic regulation, the relationship between the structure and function of proteins that enable cells to contract and maintain structure, and the way that these change under certain conditions. The research in his laboratory is focused on the proteins troponin and calponin, both of which play an important role in muscle contraction and cell motility.

One key finding that Dr. Jin’s laboratory first discovered was that TNNT1 myopathy affects very specific types of muscle cells. Our muscles have two types of fibers: “slow-twitch” muscle fibers that provide us with muscle endurance, longer-lasting energy, and the ability to move against gravity, as well as “fast-twitch” muscle fibers, the muscle fibers that give

us sudden bursts of energy but which tire easily. TNNT1 myopathy patients have normal troponin T in their fast-twitch fibers but have none of the necessary troponin T protein in their slow-twitch muscle cells. In TNNT1 myopathy patients, the complete absence of troponin T protein in the slow-twitch muscle fibers cause the large muscles that affect movement (like the shoulders and thighs) and breathing (the chest muscles) to become progressively weaker.

Further studies by Dr. Jin’s laboratory demonstrated the critical role of the troponin T in slow-twitch muscle to resist fatigue. This explains why we see severe effects on the muscles in TNNT1 myopathy patients despite normal troponin T in the fast-twitch muscle fibers (for reference, see page 4). Dr. Jin’s more recent work has focused on how the slow form of troponin T function in muscle spindles, the sensory receptors that inform the brain and spinal cord about changes in the length of individual muscles and the speed of stretching. With this information, our brain computes the position and movement of our arms and legs in space, which is needed for muscle control, posture, and a stable gait. The loss of the slow form of troponin T in TNNT1 myopathy causes features of the disease like tremors and muscle weakness. As Dr. Jin explains, “This finding sheds light on a novel treatment approach by targeted supplement of slow troponin T protein in spindle intrafusal fibers.”

Dr. Jin’s work in understanding TNNT1 myopathy is critical for developing a targeted treatment, like gene therapy, for TNNT1 myopathy patients. The knowledge of how troponin functions or doesn’t function in patients with TNNT1 myopathy lays the necessary foundation for determining the best mechanisms for treating patients- not only the Amish, but others all over the world. His TNNT1 myopathy studies have increased the clinical awareness of TNNT1 myopathy everywhere. Five other genetic abnormalities in the *TNNT1* gene have now been identified to cause similar disease to the Amish in various ethnic populations.

In his free time, Dr. Jin enjoys traveling the world, especially to Tibet, Utqiagvik (Barrow) Alaska, and Antarctica. He also enjoys cooking and preparing authentic Chinese dishes. Dr. Jin has been an instrumental collaborator of the Clinic for decades, and we are so grateful for his tireless dedication to further our understanding of TNNT1 myopathy.

2021 AMBASSADORS

We're excited to introduce our 2021 Clinic for Special Children ambassadors! A new program this year, our ambassadors will tell their stories and represent the Clinic in a variety of ways. We're thankful to each ambassador for helping us spread the word about the work of the Clinic!



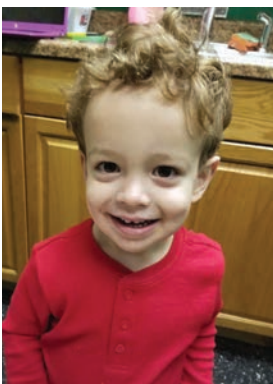
Michael Fondacaro | 23 years old *Glutaric acidemia type 1 (GA-1)*

A motivational speaker and entrepreneur, Michael is an inspiring young adult. He was diagnosed with Glutaric Acidemia type 1 (GA-1) at eight months old by a doctor who hadn't previously seen children with GA-1 survive past age three. Michael's family did their own research and found the Clinic. Michael now visits the Clinic about once a year where we monitor his condition. He has even participated in on-going research for GA-1. In his free time, Michael is busy running his motivational speaking business called 'Beyond the Chair'.



Kyreece Martin | 2 years old *Maple Syrup Urine Disease (MSUD)*

Kyreece is an adorable and outgoing toddler who loves to give smiles to everyone he meets! He was diagnosed on his first day of life with Maple Syrup Urine Disease (MSUD). He recently underwent liver transplantation at the UPMC Children's Hospital of Pittsburgh and is doing well. Kyreece visits the Clinic about every month or so for ongoing management of his MSUD. Our entire team enjoys seeing Kyreece and his family when they visit us!



Eric Rodriguez | 2 years old *Glutaric acidemia type 1 (GA-1)*

Eric is a "force of nature" as described by his parents. Born prematurely at 31 weeks, Eric received his diagnosis of GA-1 within days after his birth. After researching about GA-1, his family learned of the Clinic and had their first consultation when he was six months old. The rest is history! The Rodriguez family visits the Clinic now every three months to monitor Eric's progress. Eric recently started day care where "he is learning to share (in theory)."

RECENTLY PUBLISHED PAPERS

Tova HersHKovitz, Alina Kurolap, Galit Tal, Tamar Paperna, Adi Mory, Jeffery Staples, **Karla W. Brigatti**, Regeneron Genetics Center; Claudia Gonzaga-Jauregui, Elena Dumin, Ann Saada, Hanna Mandel, Hagit Baris Feldman.

A recurring NFS1 pathogenic variant causes a mitochondrial disorder with variable intra-familial patient outcomes.

Molecular Genetics and Metabolism Reports. 2020 Dec 30; 26:100699. doi: 10.1016/j.ymgmr.2020.100699. eCollection 2021 Mar.

Patrick J McKiernan, James E Squires, Robert H Squires, Jerry Vockley, George V Mazariegos, Kyle Soltys, Armando Ganoza, **Kevin Strauss**, Ajai Khanna, Rakesh Sindhi.

Liver transplant for inherited metabolic disease among siblings.

Clinical Transplantation. 2020 Nov; 34(11):e14090. doi: 10.1111/ctr.14090. Epub 2020 Oct 5

Olivia Wenger, Miralides Brown, Brandon Smith, Devyani Chowdhury, Andrew H Crosby, Emma L Baple, Mark Yoder, William Laxen, Silvia Tortorelli, **Kevin A Strauss**.

Biochemical phenotype and its relationship to treatment in 16 individuals with PCCB c.1606A>G (p.Asn536Asp) variant propionic acidemia.

Molecular Genetics and Metabolism. 2020 Nov; 131(3):316-324. doi: 10.1016/j.ymgme.2020.09006. Epub 2020 Oct 3.

*bolded names indicate current CSC staff authors

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A Legacy of Service and Care for Others *Janet C. Scala Patient Access Fund*

Service and a deep desire to care for others was central to every season of Janet Scala's life. In her teens, she volunteered as a candy striper at a local hospital. As a young adult, it was at the core of her decision to enroll in the University of Rochester's School of Nursing. She would meet her future husband, Dr. Robert Scala, while caring for him during a hospital stay when Robert was a graduate student at the same institution.

Together, Janet and Robert would go on to have four children. As a mother, Janet served her children's school systems as President of the elementary, middle, and high school PTAs. Janet also broke down barriers as the first woman of her party elected to her Town Council in Fanwood, New Jersey. When it came time for her to return to nursing, she created a networking group to help other women who wanted to return to their careers after raising their children.

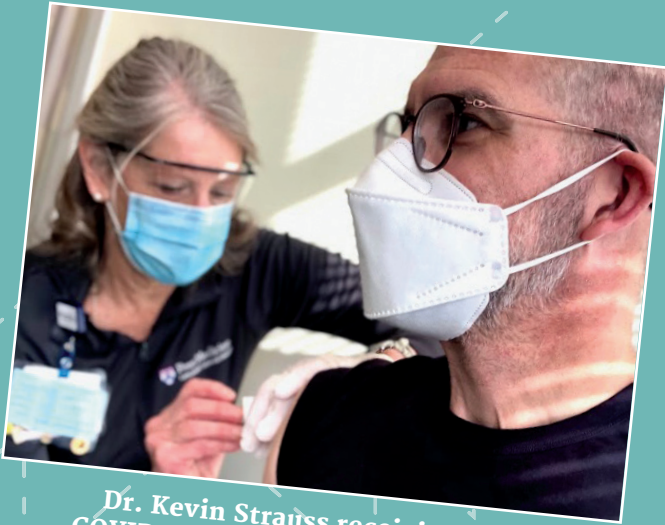
When Janet passed away in August, Robert and his children chose to honor her legacy of service and care for others by creating the Janet C. Scala Patient Access Fund at the Clinic for Special Children. Janet is remembered by her family, friends, and community as someone who had tremendous compassion for those in need. Her legacy now lives on in the Patient Access Fund, which provides Clinic patients and their families in need with vital support to offset the cost of travel to the Clinic, lodging during their treatment, and medical care received as part of their time at CSC. Because of Janet, patients and families can focus on what really matters – getting the care they need, when they need it.

The Clinic community is deeply grateful to the Scala Family for their support, and looks forward to honoring Janet's memory through offering this vital service to CSC patients.

If you are interested in supporting the Patient Access Fund or honoring the memory of a loved one, please contact Emily Seitz, Development Director, at 717-687-9407 or eseitz@clinicforspecialchildren.org.

AROUND THE CLINIC

A gallery featuring recent Clinic happenings!



Dr. Kevin Strauss receiving his COVID-19 vaccine earlier this year!



Dr. Vincent Carson receives his COVID-19 vaccine with a smile!



We had several unexpected Saturday visitors at the Clinic! Can you spot them?



Cookies for Caregivers, a local group in Lancaster, dropped off delicious cookies for our staff to enjoy!



Thank you to everyone who helped us raise over \$55,000 during the 2020 Extraordinary Give!



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2021 Benefit Auctions

for the Clinic for Special Children

See inside for more details!

June

5 Union County, PA

19 Lancaster County, PA

26 Shippensburg, PA

July

10 Shiloh, OH

August

21 Memphis, MO

September

11 Blair County, PA

We recognize the rapidly evolving situation with COVID-19 and have been taking all precautions to keep our staff and patient families safe. Auctions are subject to change due to COVID-19 restrictions.

The Clinic's Mission

"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."