DDC clinic
Center for Special Needs Children

Fall 2020
Chronicle

Enhancing the quality of life for people with special needs caused by rare genetic disorders.

When Your Child Has PKD

A Mother's Story

Isaac is our first and only child. He was born weighing a healthy 7 pounds 2 ounces and at first he was a perfectly healthy baby, or so we thought. A few hours after birth we noticed he was looking a little jaundiced, which we thought wasn't unusual since many newborns can be for a little while, but his color kept getting more and more golden to the point where medical staff didn't want to wait to check the bilirubin, so they did a blood test and it turned out to be extremely high. He was immediately transferred to University Hospitals Rainbows Babies & Children's Hospital where he was put under phototherapy lights.

We of course had so many questions, but they really couldn't give us answers as to why this was happening. Nothing was making sense. We just knew that he was extremely anemic and would probably need a blood transfusion. After a while the lights were helping the bilirubin come down. Further testing was done but we were told we wouldn't get the results for a while. He ended up needing a blood transfusion at 4 days old. He stayed under the lights in NICU for a week; after that we were able to go home. The evening before we left they told us the earlier blood tests came back, and they were pretty sure he has an inherited blood disorder called Pyruvate Kinase Deficiency (PKD). We'd never heard of this before since it's a rare disorder. We searched the internet for research on PKD, but we didn't find a whole lot. We brought him home with many follow-up checkups at his pediatrician's office and Rainbow Babies & Children's over the following weeks. A month later, genetic testing confirmed the diagnosis of PKD. Isaac continued needing blood transfusions every 4 weeks at that time. When he was about 6 months old, my mom urged us to see Dr. Wang at DDC Clinic, and we learned he had a few older patients with this disorder. Without hesitation, we took him there. DDC Clinic exceeded our expectations with the help and care they give. We also discovered this

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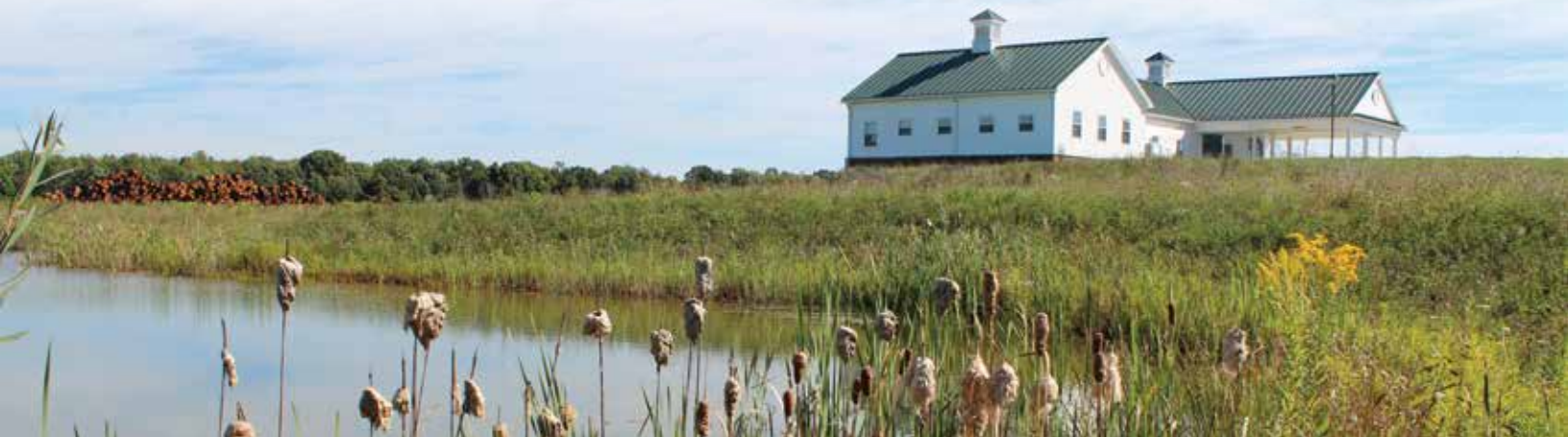

About DDC Clinic

DDC Clinic – Center for Special Needs Children is a non-profit primary care and research facility serving patients with complex medical needs. Located in Middlefield, Ohio, we have been recognized internationally for our state-of-the-art genetic research impacting broad-based health concerns such as heart disease, cancer and neurological disorders.

Our mission is to enhance the quality of life for people with special needs caused by rare genetic disorders. Founded in 1998 by five local Amish families committed to helping tomorrow's special children, we now serve over 1,000 patient families in 30 states and several foreign countries.

Our clinic is a unique collaboration of the Amish and non-Amish communities, dedicated doctors and researchers all working together to find answers for parents and treatments for children. The clinic was built on the 'medical home' concept defined by the American Academy of Pediatrics. We provide comprehensive and personalized medical care for special children with over 160 different rare conditions.

Our in-house certified clinical genetics laboratory provides rapid, high quality and affordable testing. We conduct patient centered research – always seeking faster diagnoses, better understanding and improved treatments for our children.

We offer personalized education and support services for patient families. No family is ever turned away based on ability to pay.

DDC Clinic is a gathering place; a place of love, compassion and caring; a place where people take the time to listen and share; a place of faith and hope.

Patchwork

2020 BENEFIT

An evening to support DDC Clinic for Special Needs Children

Thank You to Our Supporters

Despite its cancellation, this year's Patchwork Benefit raised nearly $50,000 for DCC Clinic, thanks to the generosity of our supporters. Patchwork, originally scheduled for April, was postponed until the fall due to COVID-19. However, with the continuing pandemic, we couldn't put our supporters at risk so we canceled the event. Many of those who had already purchased tickets or sponsorships graciously donated those dollars to our clinic. We offer our sincere gratitude and appreciation to these 2020 Patchwork donors.

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A Story of Heartache, Hope and Healing

Connor Teare was less than a year old when his parents noticed he wasn't crawling. When it became apparent he was experiencing a variety of motor skills challenges, mom Cynthia began looking for answers, answers that turned out to be difficult to find. Visits to area hospitals led to misdiagnosis after misdiagnosis, including Muscular Dystrophy and Parkinson's. By 4 years old, the Geauga County youngster had digressed from a walker to wheelchair, only able to attend Burton Elementary with the help of a fulltime aide. While his classmates were wonderfully supportive, Connor knew he was different and didn't know if he'd ever be like the other kids. Well-meaning doctors did their best to help, but just didn't have answers. Unable to determine what was wrong with their son, the Teares faced frustration and fear. Connor began to dread a lifetime of limitations and 24/7 care, struggling to maintain his spirits. His only escape was Saturdays at Fieldstone Farm Therapeutic Riding Center in Bainbridge, where, secured to a horse with a special device, he enjoyed riding and the company of other brave boys and girls.

An appointment with Dr. Heng Wang at the DDC Clinic marked the beginning of intensive research, compassionate care and, most importantly, a path to an answer. Dr. Wang's genetic testing revealed that Cynthia had a deleted gene that, passed on to Connor, reduced the flow of dopamine and all but eliminated his ability to move. Dr. Wang's prognosis, Dopa Responsive Dystonia, was passed on to Dr. Irwin Jacobs at University Hospitals, who prescribed Sinemet (Carbidopa Levodopa). Almost immediately Connor was able to pull himself up on to the bed and progressed from wheelchair to walker. By 4th grade, he shocked and thrilled students and teachers by walking into the building on the first day of school! During Connor's first year of high school, he became fully mobile, finally able to enjoy all that comes with being just another teenager.

It shouldn't come as a surprise that Connor chose a career in nursing. Graduating from a very competitive program at the University of Akron, he's in his third year as a nurse at Western Reserve Hospital. Connor recalls the care and compassion he received at DDC Clinic every time he connects with a patient, sharing with them the importance of never giving up. He resides in Macedonia with his girlfriend, and like myriad young couples, looks confidently to a future of freedom and hope.

"The DDC Clinic is a calming, loving, caring place," Connor says. "They put your best interests first, always. They are family!"

Thank You to our Auction Committees and Supporters!

Once again this year, our friends in several Amish and Mennonite communities held benefit auctions in support of our work with special children.

The 14th Annual Shiloh Mennonite Community Benefit Auction was on Saturday, July 11th in Shiloh, OH and the 8th Annual Spartansburg Benefit Auction was on Friday, August 14th in Spartansburg, PA. Blessed with beautiful weather and large crowds, the Shiloh auction raised $42,000 and the Spartansburg auction raised $69,000 for DDC Clinic.

Our last auction of the season was the 20th Annual Geauga Benefit Auction on October 10th. The largest of the three auctions, this event featured a large array of auction items including handmade quilts, furniture, tools, horses, buggies, firewood, a 24' x 32' garage and a timber frame pavilion. The auction raised over $300,000 in support of DDC Clinic.

We're very grateful to each of the communities who hold these important benefit auctions and the many community members and volunteers who lend a hand to make these auctions possible. We cannot thank these community members enough for their generosity and their donation of time, goods and labor.

The money raised from our community benefit auctions allows our clinic to provide life-changing diagnoses and treatments for special needs children and their families.

Groups and Programs Update

COVID-19 has challenged us in many ways and disrupted so many of our regular activities. Here at DDC Clinic, we temporarily suspended our immunization clinics, Mother's Coffee Break and Women's Quilting and Craft Club. Slowly and cautiously, however, we have resumed these activities and programs.

To keep everyone healthy and safe, we have protocols in place for all visitors and groups coming to our clinic, including masks, distancing and hand washing or use of hand sanitizer.

IMMUNIZATION CLINICS

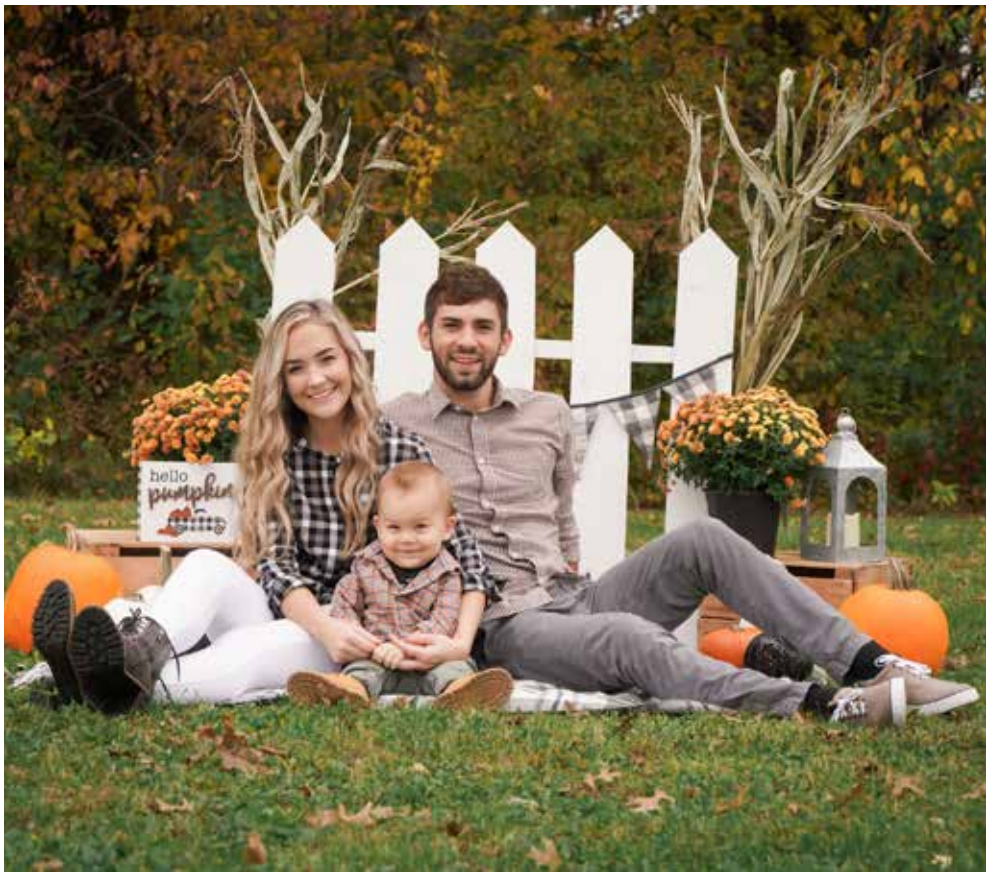
In early May, we resumed our immunization clinics as one of the first in the state, knowing that unvaccinated children are more susceptible to other serious diseases. DDC Clinic immunization clinics are by appointment only. Any child can be seen; they do not need to be a patient of DDC Clinic. Dates fill up very quickly. Appointments are available now for the month of April 2021. Since resuming in May, we've given over 648 immunizations. To schedule an appointment, call 440-632-1668.

MOTHER'S COFFEE BREAK

Our Mother's Coffee Break group began again in October. This group for mothers of special needs children meets monthly to share information, exchange ideas and visit. Occasionally, there are guest speakers. The group meets at DDC Clinic on the second Wednesday of each month, September through April from 9:30 a.m. to noon. Babysitting is not provided, but all are welcome.

QUILTING AND CRAFT GROUP

The ladies of the Quilting and Craft Group began meeting again in November. The group comes together on the first Wednesday of each month from 9 a.m. to 1 p.m. to quilt, craft and socialize. Each year, a few of the quilts are auctioned off at our Patchwork Benefit and at the Geauga Benefit Auction. The group invites any lady in the community to join.



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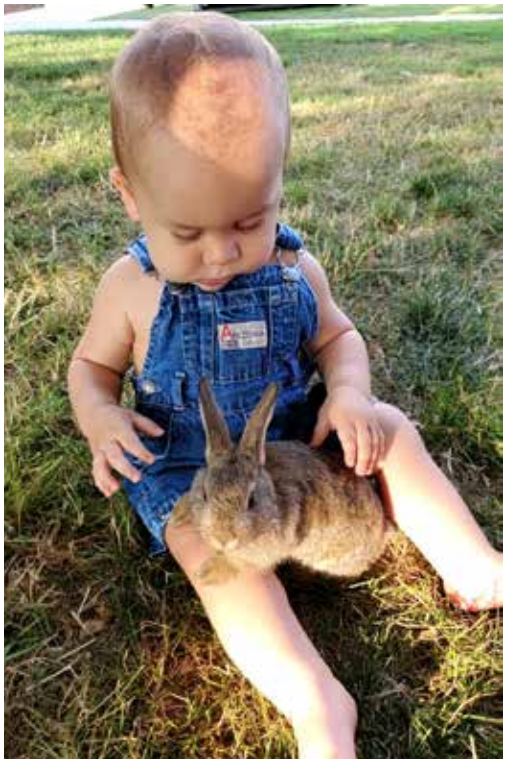
is where they performed the genetic testing we had done at Rainbow Babies & Children's Hospital. We got into a routine of blood work every 3 weeks to check his hemoglobin at DDC Clinic and a transfusion every month at Rainbow. After a while we could tell when he needed a transfusion; Isaac would start looking jaundiced and was more tired than usual. Dr. Wang informed us of what to expect and watch for: increased heart rate, iron overload and enlarged spleen. A splenectomy could also be an option in the future to slow down the need for transfusions or stop them altogether. We are so grateful to have DDC Clinic so close to home. Isaac is now 18 months old; he is otherwise a healthy, very active toddler. He's had 15 transfusions so far; he needs them about every 6 weeks now. Between regular visits at DDC Clinic and hematology at Rainbow, we can manage to keep his hemoglobin stable.



I'd like to give a special thank you to everyone who donates blood; you're doing more than you can ever imagine. With you, with God, family and the continued help and research from DDC Clinic, we believe Isaac can continue to grow strong, healthy and have a good future.

– Isaac's proud Mom, Sara

Meet Duncan, Virginia and Oliver

Duncan Tanner, Jr., our new Director of Advancement, joined our team in late September. He brings 35 years of experience in non-profit fundraising, marketing and volunteer development. Charged with many tasks essential to securing financial resources and telling our clinic's story, Duncan just completed a development plan and case statement.

"I'm excited about the opportunity to engage donors, businesses, foundations and organizations," says Duncan, "and I believe DDC Clinic's groundbreaking rare genetic disorders research and compassionate, dedicated work with special needs children will attract support from both Amish and non-Amish communities."

He resides in Stow with his Rottweiler, Angus, and he enjoys travel, going to the gym and cheering for the Browns.

Our new administrative assistant, Virginia Bell, joined DDC Clinic in May 2020. She is the first person you see as you enter DDC Clinic and the caring voice that answers the phone. She has lived and worked in the Middlefield area for more than 40 years. Experienced in sales and customer service, she most recently worked for a physical therapy office.

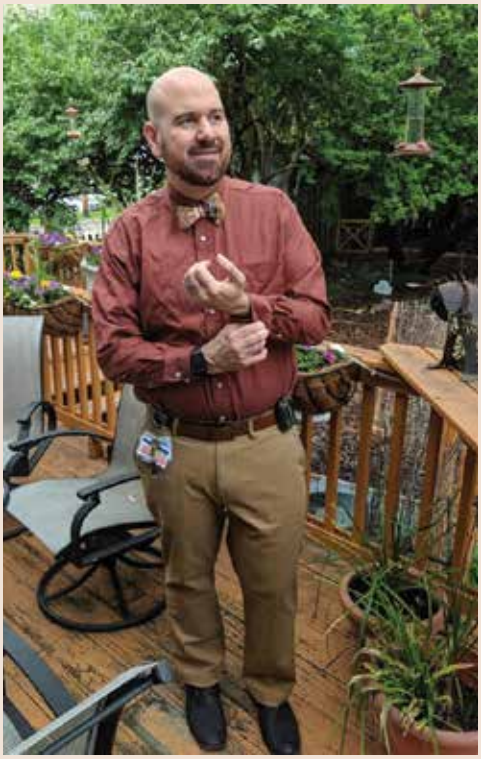
"I'm very happy to be working and giving back to the community I live in," says Virginia.

Virginia enjoys spending time with her family, mowing the grass, and collecting beach glass from the shores of Lake Erie. She is also known for her love (obsession) with decorating Christmas Trees. She decorates over 50 trees in her house every year.

Oliver McCourt recently joined our team as a full-time lab technician. He interned with DDC Clinic for two months this past summer and was offered his position upon completion of his internship.

"I met some incredibly talented and passionate people along the way. I couldn't imagine a more professional and hands-on learning experience," says Oliver. "I've been humbled by this opportunity and I'm happy to continue working with DDC Clinic."

Say Hello to Dr. Cruz



DDC Clinic is pleased to announce that Dr. Vincent Cruz will be spending the next year at DDC Clinic as he finishes his physician training in Internal Medicine and Genetics.

Dr. Cruz is one of the KeyBank Minority Medical Student Scholars of the Cleveland Clinic Lerner College of Medicine of Case Western Reserve University.

He will focus on research projects while at DDC Clinic and will graduate in May of 2021. He considers himself a physician scientist and looks forward to exploring the application of genomics to individualized patient care with Dr. Wang and Dr. Xin.

Dr. Cruz received his Doctor of Medicine from the Cleveland Clinic Lerner College of Medicine and Case Western Reserve University in 2016. Originally from Detroit, Dr. Cruz is married and has three children.

He enjoys fly fishing and astronomy. He is exploring bicycle riding and looks forward to trying some of the local bike paths. His wife is currently enrolled in a PhD program through Case Western Reserve University to study how genomics affects large populations.

Dr. Cruz got to know Dr. Wang over the last three years through consultations about unusual and challenging patients. He found Dr. Wang to be very responsive and collaborative, which is why he is here. He said Dr. Wang is well respected at Case Western Reserve University. He shared that he recently had a patient who was tested for Cohen Syndrome by a different laboratory. The report came back from the lab confirming the patient was a carrier of Cohen Syndrome and two of Dr. Wang's publications were cited by the laboratory.

Dr. Cruz is attracted to DDC Clinic's rural setting and more importantly, the direct impact DDC Clinic has on individuals and the community. He wants to study undiagnosed diseases and find answers to what is unknown. He especially wants to help families find answers about the rare conditions affecting their children.

Welcome to the DDC Clinic family, Dr. Cruz!

Understanding Genetics

Dr. Vincent Cruz, MD, MS, BSEE is chief resident and research fellow training in Internal Medicine and Medical Genetics at Case Western Reserve University and Rainbow Babies / University Hospitals Cleveland Medical Center's Center for Human Genetics, spending his last year of training at DDC Clinic studying genetic diseases. He shares his knowledge about PKD below.

Q: What is Pyruvate kinase deficiency (PKD)?

A: PKD is a "rare" genetic blood disorder that poses specific challenges at different times in life for affected individuals. It causes red blood cells to be more fragile than normal resulting in specific problems throughout a person's lifetime. PKD affects individuals from around the world and from many different backgrounds, but people from Europe and China seem to be affected more often. Some estimate that 1 out of 100 people may be an unaffected "silent carrier" of this disease, but we really don't know the true number. Within the Plain Community, PKD is found much more frequently because there are many more silent carriers than in the general population.

Q: What happens to people with PKD?

A: There are different concerns at different stages of life for individuals affected by PKD. Most of the complications of PKD come from increased fragility of the red blood cells (RBCs) in affected individuals. RBCs are small cells that make our blood red-colored, and contain special proteins including hemoglobin that deliver oxygen to our body. Having fragile RBCs means that these packets of hemoglobin and other proteins are more likely to break apart when traveling around the body. Because of this, RBCs in people with PKD tend to wear out very quickly. This causes anemia but also the content of the broken RBCs spills into the bloodstream and can damage certain organs in the body.

Q: What are those times of life that having PKD is most concerning?

Although it's a life-long disease, the most concerning time for a person with PKD is early in life. Some PKD-affected babies during pregnancy can develop too much fluid. This is a serious and life-threatening, but uncommon, condition. Probably most concerning is the breakdown of the contents of the RBCs as a baby is starting to breathe on their own for the first time. This can cause "hyperbilirubinemia" – a build-up of toxic substances from the breakdown of the RBC contents – and "jaundice" or yellowing of the skin and eyes in the first few days or weeks of life. If untreated, this can irreversibly affect the brain and be life-threatening.

Challenges after birth can include poor growth and lethargy. In times of stress, children and adults with PKD may break down their RBCs faster and may become dangerously anemic, requiring blood transfusions. This is especially true for certain viral infections like parvovirus and during pregnancy in women affected by PKD.

The constant breakdown of RBCs can swell the spleen (an organ used to filter the RBCs) so much that it can become uncomfortable and need to be surgically removed – which can make individuals susceptible to certain infections. They can also develop gallstones. It is important for people with PKD to have regular check-ups with their doctors to monitor for these and other complications.

Q: How do you inherit PKD?

A: PKD is an "autosomal recessive" disease caused by misspellings in a gene called PKLR. Think of genes as the "recipes" used by our bodies to make us who we are. There are over 20,000 genes in each of us, and they come in chromosomes (or "cookbooks") passed down to us from our parents. Many genes are for components of your blood including PKLR. Everyone has two copies of PKLR – one passed down from each parent. You can think of the second copy of PKLR as a "backup" in case there is a misspelling in the first copy. Just like you only need one correct recipe to make dinner, your body only needs one correct copy of PKLR to be healthy. In "autosomal recessive" conditions like PKD, if BOTH copies of the gene have a misspelling, you can't properly make the components of the blood.

Q: Why might I want to know if I am a carrier?

A: Many families may not know that Mom and Dad are both carriers of PKD until they have a child diagnosed with the disorder. PKD can be very serious in the newborn period, so it is very helpful to be "on the lookout" for signs of PKD and treat it much quicker. When BOTH you and your spouse are known carriers, there is a 1-in-4 chance that each of your children might have PKD, although there is no way to predict how many children will actually be affected.

DDC Clinic Receives Grant from Midwest Genetics Network

DDC Clinic recently received a grant from Midwest Genetics Network supporting our educational effort on newborn screening of rare genetic conditions in the Amish Community.

Families of children with rare conditions frequently experience a diagnostic odyssey trying to find out what's wrong with their child. Not only is this delay in diagnosis and treatment detrimental to the child, but the family and child frequently experience hospitalizations which are expensive and frustrating.

DDC Clinic has developed an Amish genetic panel test which can diagnose 160 rare conditions and provide answers to families. Through our partnership with Middlefield Care Center, our clinic provides the genetic panel test to newborns, resulting in earlier diagnoses and life-changing intervention.

The grant from Midwest Genetics Network has made it possible for Carol Troyer Counselman, DDC Clinic RN, to have one-on-one educational meetings with families of newborns to share important information about the genetic panel test.

My Unique Experience at DDC Clinic

My name is Dr. Lauren Pronman. As part of my residency training in Pediatric/Genetics, I was able to spend some time at DDC Clinic – Center for Special Needs Children. This was both a unique and educational experience. My goal while rotating through this clinic was to learn more about different rare disorders, which I did successfully accomplish. However, I quickly realized that my time at DDC would be different than the usual physician-patient interaction; instead, it was a place where the families taught us just as much as we taught them. Dr. Wang showed me how to learn through your patients about these rare conditions and how patient lifestyles may change the way you practice medicine. He showed me that sometimes the gold standard of management and/or diagnosis may not be the best option for the family sitting in front of me. In addition, my time on this rotation allowed me to get a glimpse into their Amish community, which in turn will make me a better resident and future physician.

DDC Clinic's Scientific Contributions to Understanding PKD

Red cell pyruvate kinase deficiency (PKD) is a rare congenital hemolytic anemia caused by genetic defects in the PKLR gene. DDC Clinic, who is emerging as a global leader on PKD, has developed research and clinical expertise in the specific medical challenges that PKD patients face at every stage of life by providing medical care to patients from newborns to older adults.

In 2014, our clinic joined Dr. Racheal Grace of Harvard Medical School and Dr. Holmes Morton of Central Pennsylvania Clinic for Special Children and Adults to establish an international, multi-center registry for PKD to collect clinical data on affected patients and further advance our understanding of this condition. Our recent natural history study on this disease has enrolled over 250 patients and has led to 5 publications in prestigious peer-reviewed medical journals:

1) *Clinical spectrum of pyruvate kinase deficiency: data from the Pyruvate Kinase Deficiency Natural History Study.* ***Blood****, 2018.*
2) *Prevalence and management of iron overload in pyruvate kinase deficiency: report from the Pyruvate Kinase Deficiency Natural History Study.* ***Haematologica****, 2019.*
3) *Characterization of the severe phenotype of pyruvate kinase deficiency.* ***American Journal of Hematology****, 2020.*
4) *The pyruvate kinase (PK) to hexokinase enzyme activity ratio and erythrocyte PK protein level in the diagnosis and phenotype of PK deficiency.* ***British Journal of Haematology****, 2020.*
5) *Genotype-phenotype correlation and molecular heterogeneity in pyruvate kinase deficiency.* ***American Journal of Hematology****, 2020.*

Three additional manuscripts are in review for publication in professional journals.

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Enhancing the quality of life for people with special needs caused by rare genetic disorders.

Together, We Can Change Children's Lives

With your help, we can make a difference in the lives of children like Michael who are affected by rare genetic disorders.

Michael's family is just one of more than 1,000 families with special needs children that we've helped over the years.

This past year, our clinic has faced new financial challenges brought on by COVID-19. We had to cancel our annual Patchwork Benefit, one of our most important fundraisers. Patient visits to our clinic have decreased, while expenses have increased. We had to purchase more medical supplies like PPE to keep our staff and patients safe. Donations from our supporters that are critical to our day-to-day operations have diminished.

Now, more than ever, we need your help. With your support, we can continue to provide the crucial medical care that children like Michael need.

Will you consider making a donation to help a child live a better, healthier life? Your gift can make a real and lasting impact on the lives of special needs children and their families.

To make a gift, please contact us at 440-632-1668 or return the donation form inside. We thank you for your continued support.