DDC clinic
Center for Special Needs Children

Chronicle
Summer 2021

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

Welcome Dr. Cruz

The Board of Directors and staff of DDC Clinic are happy to announce Dr. Vince Cruz will be joining the clinic on July 1, 2021.

Dr. Vincent Cruz and Dr. Heng Wang

Dr. Cruz was trained in both Genetics and Internal Medicine. His specialties allow DDC Clinic to move forward with continuity of comprehensive care. Dr. Heng Wang, Medical Director of DDC Clinic stated, "Dr. Cruz joining us will allow us to comfortably follow our patients as they age into adulthood."

Dr. Cruz is one of the KeyBank Minority Medical Student Scholars of the Cleveland Clinic Lerner College of Medicine of Case Western Reserve University and received his Doctor of Medicine degree from the Cleveland Clinic Lerner College of Medicine and Case Western Reserve University in 2016. Dr. Cruz completed his residency at University Hospitals Genetics Department and a one-year research fellowship at DDC Clinic.

DDC Clinic's patient base, research needs and professional collaborations have grown steadily over the years. Since hiring our first physician and medical director, Dr. Heng Wang, in 2002, the number of patient families we have served has grown to over 1,500. By providing diagnostics, consultations and medical education to other physicians, clinics, hospitals and research centers across the country and around the world, we have impacted thousands more.

The clinic's growth and expanding collaborations have created opportunities for new discoveries, treatments and cures. A second physician allows us to take what we have learned from our research and apply it to new therapies and treatments, advancing highly personalized care that is the core of our mission.

The generous support of many made it possible for DDC Clinic to hire our second physician. Dr. Cruz's work will contribute to the services we provide all of our patients. Please consider a gift to our Physician's Fund to support his work at DDC Clinic.

Reflections

By Dr. Heng Wang

When I recently recounted the story of my first trip to Middlefield, I realized that it had been almost 20 years since the clinic opened its doors for special needs children in 2002.

It has been such a humbling but privileged experience to serve thousands of children and families affected by rare genetic diseases, locally, nationally and internationally. The clinic has grown from a solo house-call doctor to a state-of-art medical facility with cutting-edge research and a highly demanded molecular diagnostic laboratory, serving patients, families and health professionals around the globe.

DDC Clinic has demonstrated a unique model in which biomedical research may integrate and synergize with

> PAGE 7

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.

Comprehensive and Continuity of Care

By Dr. Vince Cruz

I'm honored to have been selected as DDC Clinic's second physician and to be part of such a dedicated and compassionate team. In my new role, I'll provide our patients with comprehensive care and continuity of care.

The terms "comprehensive care" and "continuity of care" may seem similar, but they have very different meanings. DDC Clinic provides comprehensive care, managing as much patient care as possible in-house, from diagnosis to therapies to research into rare diseases. Our comprehensive care has saved families in our community from traveling to different doctors and facilities to get the same quality and types of medical services.

Continuity of care is the concept where a team of doctors and nurses actively work together to provide all the care a patient needs over time as they mature. Our adolescent patients with special needs are growing up and becoming young adults. To continue to serve these patients, we have widened our clinic's scope of practice to include continuity of care.

Over the past two decades, our clinic has greatly expanded our "breadth" of service by offering vaccinations, newborn screening and access to research. Now, by adding continuity of care, we're expanding our "depth" to include older individuals with special needs.

As second physician, I'll help to provide quality care to our patients. It's important for me to state that I am not here to replace Dr. Wang. I bring a different set of skills and plan to share the workload. We hope to provide a seamless transition of care for families as their children get older and grow to adulthood. We plan to make DDC Clinic a "lifelong medical home" for all those in the Plain Community who are affected by rare disease, regardless of age.

I look forward to extending our comprehensive care to adults in our community, and I'm eager to meet you – our patient families – and hear your ideas about how we can better serve your needs.

Understanding Genetics

Genetic Medicine in Adults

Most people think of genetic diseases as affecting “only children,” expecting pediatricians to be interested in genetics, but not someone trained in internal medicine. The fact is that most geneticists come from a pediatric background.

It makes sense that pediatricians are interested in genetics as many children are affected by genetic conditions. Pediatrics is where the field of clinical genetics first focused. As we learn more and can do more about genetic conditions, it has become a broader field that extends naturally into adult medicine for a couple of reasons.

First, children with genetic conditions are living longer, in part due to amazing research and comprehensive care from physicians like Dr. Xin and Dr. Wang. In the past, these children may have not lived long enough to develop unrelated adult diseases or long-term complications of their genetic disorders. As time goes by, the number of adults affected by pediatric genetic conditions will continue to grow.

Second, we’re learning that many diseases of adults are at least in part influenced by genetics. Nearly half of all diseases are now thought to have a genetic component or risk factors. Heart disease, diabetes and dementia are all “hot topics” for genetic researchers around the world.

With mounting evidence of how genetics affects disease throughout a person’s life, it’s inaccurate to say that genetic conditions only affect children. It’s not a matter of “if” but “when” adult medicine doctors will have to manage their patients’ genetic diseases, and our clinic intends to be at the forefront of this new frontier in medicine.

Reflection from Dr. Cruz on his decision to join DDC Clinic:

“When I was asked, ‘Why study genetics and internal medicine?’ I carefully explained that there are still many unknowns in how genetics plays a role in adult medicine. I’m excited by the unknown, and to be trained in genetics and adult medicine is a blessing because it gives me tools and insights into an area of medicine that has not been fully investigated.

DDC Clinic and Dr. Wang are giving me the opportunity to apply the tools of my training to real-world problems of the Plain Community – truly a dream career for me. I look forward to being productive and exploring the long-term consequences of rare disorders as our patients age, while simultaneously providing continuity of care as their changing needs evolve.”

Recent Publications

Patient-oriented research advances diagnoses and ground-breaking treatments, bringing relief to children and hope to their families. Sharing what we learn in serving children with special needs benefits all the world’s children.

“From disease description and gene discovery to functional cell pathway: a decade-long journey for TMCO1.” *Frontiers in Genetics*, April 2021

“Tissue-Biased Expansion of DNMT3A-Mutant Clones in a Mosaic Individual is Associated with Conserved Epigenetic Erosion.” *Cell Stem Cell*, August 2020

“Characterization of the Severe Phenotype of Pyruvate Kinase Deficiency.” *American Journal of Hematology*, July 2020

“Genotype-phenotype Correlation and Molecular Heterogeneity in Pyruvate Kinase Deficiency.” *American Journal of Hematology,* May 2020

“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*, May 2020

“A Male Patient Inherited a Novel GCH1 Deletion from an Asymptomatic Mother.” *Journal of Movement Disorders*, May 2020

“Proteomic investigations of human HERC2 mutants: Insights into the pathobiology of a neurodevelopmental disorder.” *Biomedical and Biophysical Research Communications,* March 2019

“Prevalence and management of iron overload in pyruvate kinase deficiency: report from the Pyruvate Kinase Deficiency Natural History Study.” *Haematologica*, 2018

“Bilateral angle closure glaucoma in a 28-year-old Cohen syndrome patient.” *Ophthalmic Genetics,* 2018

“Oral ganglioside supplement improves growth and development in patients with Ganglioside GM3 Synthase Deficiency.” *JIMD Reports*, 2018

“The clinical spectrum of Pyruvate Kinase Deficiency: Data from the Pyruvate Kinase Deficiency natural history study.” *Blood*, 2018

*To view a complete list of publications, please go to ddcclinic.org and select **Research**.*

Donation of Love

The family of Linda Mast wanted to remember their daughter Linda in a special way. They contacted the Cohen Syndrome Association to make a donation. The Association contacted DDC Clinic to use the funds to have a poly-vinyl bench made with an engraved plaque to always remember Linda. DDC Clinic is very proud to have this bench at our facility and most thankful to the Mast family and The Cohen Syndrome Association. What a wonderful way to remember a special girl.

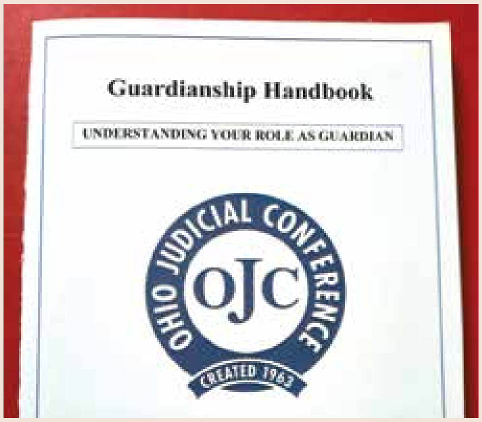
Guardianship Questions Answered

As a child with disabilities nears his or her 18th birthday, parents often wonder whether they should seek a guardianship over their child. The short answer to this question is, "it depends." In most states, a parent is deemed to be the legal guardian of his or her child until the child turns 18. Until 18, parents have the legal authority to make decisions (medical, financial, etc.) for their child. Most providers of services, including physicians, dentists, and school personnel, do not question this authority when the parent is in charge of his or her minor child and the parent is making decisions, recommendations, and participating in all of the areas where a child needs to be represented. The minute the child turns 18 years of age that authority ceases. The parent must then decide whether to seek decision-making authority for the child, and if so, how much authority. The person given the authority to make decisions is called a guardian.

A guardian is appointed by the probate court for an incapacitated person (sometimes called a "ward" or "respondent") and the guardian can be in charge of some or all personal affairs of the incapacitated person.

Not every child who has disabilities needs to have a guardian. If the child is able to make good decisions, then he or she may not require a guardian or conservator at all.

Who may be a guardian? Any person 18 years of age or older may be a guardian; the harder question is who should be the guardian. Often parents will petition the probate court to be the guardians of their child and usually the petition is granted. Sometimes the court will appoint one parent as guardian, other times both parents will be appointed as co-guardians. In some cases where the parents of the child do not live together and cannot agree on who should be appointed as guardian, the proceedings may become contested, and the court will appoint an independent guardian.



Guardianship Handbook

UNDERSTANDING YOUR ROLE AS GUARDIAN

OHIO JUDICIAL CONFERENCE

OJC

CREATED 1963

Parents should also take steps to name a successor guardian to serve after the parents have both died or are no longer able to care for the child.

Please call Patti Gallagher at DDC Clinic with questions about guardianship.

Therapeutic Phlebotomy – An Affordable Adult Service

For the last four years, DDC Clinic has been running a Therapeutic Phlebotomy program to treat several iron overload conditions by Valerie Sency, RN.

Valerie Sency, RN

One of the contributing reasons for the Therapeutic Phlebotomy service at DDC Clinic is the number of patients in our Plain community that have been diagnosed with hemochromatosis, pyruvate kinase deficiency (PKD) or spherocytosis. As these diseases progress, excess iron builds up and starts to damage essential body organs. Removal of a large amount of blood through the therapeutic phlebotomy, similar to blood donation, is a simple, but very effective treatment in these iron overload conditions.

Since a registered nurse is managing the service of blood removal through the therapeutic phlebotomy, the service is convenient and affordable. By offering this service at the DDC Clinic, patients do not need to see a specialist or travel far to a different location. All can be handled at DDC Clinic. The cost of the service is only a small fraction of what it costs at major medical centers.

During the visits, Valerie performs a blood removal, takes a blood sample to monitor hemoglobin and ferritin levels and ensuring the patient doesn't become overly anemic. While meeting with patients, Valerie also discusses other wellness options, like foods and supplements to avoid and reviews previous lab results, creating a holistic approach to care.

Iron overload conditions are detectable in children and adults. Successful treatment and management are dependent on early detection. DDC Clinic is dedicated to providing affordable treatment and management for adults with our Therapeutic Phlebotomy service.

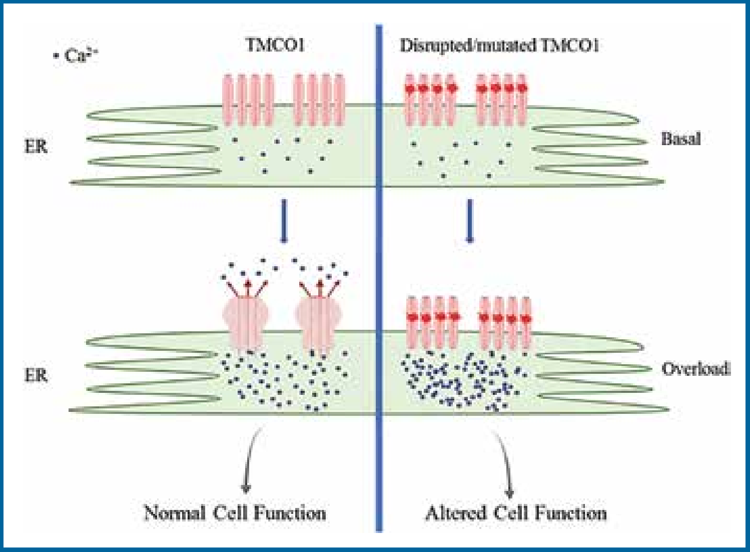
TMCO1 Defect Syndrome Published in *Frontier in Genetics*

DDC Clinic's ongoing work on TMCO1 Defect Syndrome "From disease description and gene discovery to functional cell pathway: a decade-long journey for TMCO1" just published in ***Frontier in Genetics***. This review, using TMCO1 Defect Syndrome as an example, has highlighted the dramatic progress in the studies of genetic disorders in the Plain populations through new technologies and methodologies in genomic medicine during the last two decades.

In 2010, we identified a novel autosomal recessive condition in a group of 11 undiagnosed patients within the Old Order Amish of Northeastern Ohio with similar clinical features, and later named it TMCO1 Defect Syndrome. Through this publication, we share our decade-long journey to understanding this novel rare genetic disorder: transmembrane and coiled-coil domains 1 (TMCO1) defect syndrome; from disease description to functional studies in both cell and animal models, and application of learning back to patient management.

This endeavor exemplifies how translational genomic medicine can benefit affected patients and families both within Plain communities and the broader international rare disease community, and demonstrates how a rare genetic disorder may help us to understand common and essential biological pathways in human cells.

https://www.frontiersin.org/articles/10.3389/fgene.2021.652400/full



DDC Clinic is grateful for the support from the following friends:

Prentiss Foundation, Elisabeth Severance
For their longtime support and most recent gift of $90,000 in operating support.

Catherine L. and Edward A. Lozick Foundation
For the $75,000 grant award for the Second Physician Scientist Fund.

Fred A. Lennon Charitable Trust
The Lennon Trust contribution has helped us maintain all the critical services that DDC Clinic offers with a grant in the amount of $75,000.

Ohio Developmental Disabilities Council
For the $50,000 grant award in continued support for targeted research and community outreach services.

Bessie Benner Metzenbaum
We are grateful for the fund's ongoing commitment to our mission and program services. Thank you for the general operating support award of $30,000.

Cleveland Foundation
Thank you to the Cleveland Foundation for their commitment to our work and their most recent award of $25,000 for The Rapid Response Fund.

Cleveland Foundation
The Leonard Krieger Fund of The Cleveland Foundation award of $20,000 for Marketing and Strategic Planning.

United Way Services
Thank you for your support of our programs with a grant in the amount of $11,244.54

Michigan Public Health Institute – Midwest Genetics Network
Thank you for the support of our genetics education program in the amount of $10,000.

Tenaya Therapeutics, Inc.
Thank you, Tenaya Therapeutics, Inc., for your gift of $5,000 in general operating support.

The Children's Guild
Thank you to the Children's Guild for their commitment to our work and their most recent award of $5,000 for the patient assistance program.

Residents Learn About Rare Genetic Disorders

DDC Clinic is proud to host residents from University Hospitals resident programs. This collaboration allows residents to learn more about rare genetic disorders in a clinical setting that specializes in them.

"As part of our Global Health rotation at Rainbow Babies and Children's residency program, we had the opportunity to spend time at DDC Clinic for Special Needs Children. Our goal during the rotation was to learn more about rare genetic disorders and the overall care that goes into taking care of the patients and their families. Dr. Wang taught us to 'think outside the box' regarding management decisions and taking into consideration the whole family picture. Dr. Wang and the rest of the staff at DDC made it an exceptional learning experience and we highly recommend it to other residents."


Dr. Michelle Pintea


Dr. Laura Sullivan


Dr. Sonia Gera, Dr. Heng Wang and Dr. Supraja Swamy

LABORATORY TECHNICIAN INTERNSHIP


Olivia DelSignore

"My time spent at the DDC Clinic for Special Needs Children has changed me for the better. Everyone in the office and lab are kind and always want to help others. The DDC Clinic is a very special place and you will always feel at home as soon as you walk in. I can't thank everyone enough for letting me work with them and for teaching me kindness, compassion, and empathy."

SAVE THE DATE

Thank You Event for all donors and supporters of DDC Clinic.

September 17th and 18th, 2021.

Come meet Dr. Cruz!

Additional Support for Patient Families

Assisting patient families with little or no cost prescriptions, medical supplies and formula has been a service DDC Clinic has offered for years. As everyone is more aware of repurposing, we often have specialized equipment or other items such as diapers, nutritional supplements, feeding supplies and bath chairs that a family wishes to donate to another special child. We organize the items and distribute to other patients or other families in the community that have a need.

Currently with a generous donation, we are able to provide a food pantry to our community. The pantry consists of canned and boxed food with a variety of protein products like pork and beans, fruits – apple sauces and peaches and vegetables like potatoes and diced tomatoes.

The new food pantry can be accessed from our front foyer Monday through Friday 9am – 5pm. We encourage anyone who can use any of these items to stop and fill a bag. If you would like to make a donation to our pantry, please contact DDC Clinic at 440-632-1668.

Reflections > from page 1

comprehensive care for our patients. This has completely changed the outcomes of affected children and families, and even been lifesaving – a great example of translational research and personalized medicine in a new era.

However, we believe that our clinic roots are right here in the Plain community. The patients and families we serve are the very reason DDC Clinic was started and will remain at the center of our future clinic. As our clinic grows older, we are mindful that the children we serve also mature and become young adults. In 2010, DDC Clinic renewed its mission "to enhance the quality of life for people with special needs caused by rare genetic disorders" to emphasize the continuity of care as these children grow up as well as the needs of adult services. Today, with great enthusiasm, we welcome Dr. Vincent Cruz to join DDC Clinic to better our mission!

Auction Season is beginning

We are grateful to the communities who hold these annual benefit auctions and the many community members and volunteers who lend a hand to make them possible. The money raised from our community benefit auctions allows us to provide life changing diagnoses and treatments for special needs children and their families.

- The Annual Shiloh Mennonite Community Benefit Auction will be held on July 10, 2021. The location of this auction is Blooming Grove Produce Auction, 1091 Free Rd, Shiloh, OH 44878. Auction time is from 7:00am to 4:00pm.
- Spartansburg, PA auction date and location is to be determined.
- Geauga Benefit Auction will be held on October 9, 2021 at Buster Miller's Farm, 17719 Newcomb Road, Middlefield, Ohio. Begin the day with breakfast at 5:30am!

DDCclinic
Center for Special Needs Children

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

5K raises $3,600 in the first Run for the Children

April 25, 2021 was a damp cool morning. Over 40 brave individuals participated in our latest fundraiser – Run for the Children a 5K run and 1 mile walk. Thank you to sponsors, UH Geauga Medical Center, Preston Superstore, Precision Orthopaedics, Kinetico and Sport Rack. The fastest time was Aaron Laning with a chip time of 19:59!