Virtual Visit Helps Los Angeles Family

COVID-19 has presented new challenges for our clinic and how we provide patient care for the children with special needs. When traditional office visits aren't possible, telehealth visits have provided a way for us to assist in the care of children like Liam, who is affected by Cohen syndrome.

A Mother’s Story
Liam is our third child. His brothers are four and seven. Liam was born full-term but weighed only five pounds (his brothers weighed eight pounds at birth). He had microcephaly, hypotonia, developmental delay and difficulty gaining weight.

During his first few months of life, we saw a neurologist, otolaryngologist, gastroenterologist and a geneticist. It took months of fighting our insurance company for them to approve Whole Exome Sequencing (WES) genetic testing.

In January, as a result of the WES, we received Liam’s Cohen syndrome diagnosis. While we have an excellent medical team in Los Angeles, none of our doctors had ever heard of Cohen syndrome. We joined a Facebook group for parents of children with Cohen syndrome, and we immediately became part of a supportive and informative international community of families affected by this rare genetic disease.

Through this group, we learned about DDC Clinic, and excitedly planned a visit to coincide with our travel to this year’s Cohen Syndrome Association Conference in Cleveland. However, the June conference was cancelled due to the COVID-19 pandemic.

While Liam is now a happy and thriving 17-month-old, we still had so many unanswered questions. We knew that Dr. Wang has seen over 100 patients with Cohen syndrome – 100 times more than the majority of our doctors in Los Angeles. (Our team now also includes a hematologist, immunologist, endocrinologist, orthopedist and retina specialist.)

Since we wouldn’t be going to Cleveland, I emailed the clinic to ask if it would be possible to set up a virtual visit with Dr. Wang. I was overjoyed when I received a warm and timely response. Within a week, DDC Clinic had set up a Zoom call with us.
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.

Meet Our New Executive Director

We’re pleased to welcome Scott Brace as our new Interim Executive Director.

A DDC Clinic board member since 2015, Scott has 45 years of experience working with individuals with developmental disabilities. He spent 13 years as superintendent of the Holmes County Training Center where half of the infants, children and adults served were Amish. Most recently, Scott served as a consultant to help start and fund the Sunshine Training Center adjacent to DDC Clinic.

Scott assumed his new role at DDC Clinic in March. In his current capacity, Scott works with Dr. Wang and Patti Gallagher on new funding and grant opportunities to further our clinic’s mission and help special children and their families.

DDC Clinic Faces Challenges of COVID-19

Dear Friends,

All of us are struggling to adapt to the new reality of living with COVID-19. Our best scientists and doctors are trying to understand the virus and seek ways to prevent and treat it. Currently, there’s no effective treatment or vaccine.

That’s why protective measures are so important. Washing your hands frequently, wearing a mask and disinfecting surfaces help to keep the virus from spreading. Although difficult, staying six feet away from others and not gathering in large groups is critical.

At DDC Clinic, we have kept our facility open for our patients. Our vulnerable children are why we’re here, and we need to protect them. We miss seeing the Ladies Quilting Group, the Coffee Club, MSI clients and staff as well as others who use our facility.

Dr. Wang has also supported families by telephone consultations and telehealth visits. Bao, Julia and Karen still perform important tests in our lab. We practice what we preach by wearing masks, washing our hands and disinfecting after every patient visit.

Our clinic is facing significant financial challenges. Our board and staff are exploring new financing options to keep our doors open and get us through this difficult time. We recognize that many of our supporters are also struggling, and we send our prayers.

For those who are able, please consider supporting DDC Clinic with a donation. Anything you can give would be truly appreciated.

Thank you and stay safe,

Scott Brace
Interim Executive Director
A rare disease is defined as a condition that affects fewer than 200,000 people in the United States. Rare diseases are not as rare as you think – more than 7,000 have been identified so far. Approximately 4% of the global population, or 300 million people, live with a rare condition. Patients and families affected by rare genetic disorders often experience incredible challenges in finding the answers for the diagnosis, treatment and social support they need.

Since our clinic’s modest founding in 1998, we have grown into a specialized, world-class medical facility focused on improving the health, welfare and potential of people with rare genetic disorders. We believe that while the condition may be rare, the diagnosis and treatment should not be.

Partnering with our local community and an ever-expanding network of worldwide collaborations, we’ve been involved in cutting-edge research which has achieved important breakthroughs and life-changing outcomes for patients with rare genetic diseases.

We’ve shared the success of our translational research with healthcare professionals and scientists around the globe by contributing to over 60 peer-reviewed scientific publications.

Over the last 20 years, our clinic has gained recognition as a leading international medical facility for many rare genetic diseases including:

- Prolidase deficiency
- Cohen syndrome
- Ganglioside GM3 synthase deficiency
- Glucose-galactose malabsorption
- TMCO1 defect syndrome
- SAMS association
- HERC2 defect syndrome
- Cockayne syndrome
- Hypertrophic cardiomyopathy
- Microcephalic osteodysplastic primordial dwarfism type I
- Pyruvate kinase deficiency
- Byler disease
- Galloway-Mowat syndrome
- Troyer syndrome
- Tatton-Brown-Rahman syndrome

Our clinic currently provides medical services for children with more than 160 different rare disorders. We’ve served 1,200 patient families from Northeast Ohio, 37 U.S. states, Canada, Europe, Australia and New Zealand.

Our reach is worldwide, yet we remain a gathering place – a place of love, compassion and caring where children and families are respected; a place where people take the time to listen and share; a place of faith and hope.

**Dr. Wang Speaks at Ohio Rare Disease Day**

Dr. Wang appeared as one of the featured keynote speakers at Ohio Rare Disease Day at the Harrington Discovery Institute at University Hospitals in Cleveland on February 27. His presentation, titled Fighting Rare Genetic Diseases – A Community Effort, focused on the role DDC Clinic plays in identifying new and rare genetic disorders.

Hosted by NORD (National Organization for Rare Disorders) and the Ohio RAN (Rare Action Network), this annual event had more than 60 people in attendance with an additional 240 watching on Facebook.

This year’s Ohio Rare Disease Day was an inspirational and educational gathering that brought together patients, their families, caregivers, medical professionals, researchers and policy makers. While raising awareness of rare diseases was the main goal, the day-long event was also an opportunity to discuss issues, share ideas and tell personal stories. One participant who shared her experiences spoke of her 40-year journey to find a team of doctors who ultimately identified her disease.

**Rare Genetic Diseases**

Not Only Our Expertise, But Also Our Passion

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Virtual Visit Helps Los Angeles Family

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Our virtual visit with Dr. Wang has been the highlight of our time in isolation. His extensive knowledge of Cohen syndrome, coupled with his kindness and genuine affection for this population, led to a productive meeting that left us feeling hopeful about Liam’s future.

Dr. Wang also offered practical advice on treatment options for Liam. He taught us about common issues affecting children with Cohen syndrome and provided helpful options and resources to augment Liam’s mental and physical potential.

Having seen so many children with Cohen syndrome, Dr. Wang was also the only physician able to tell us whether Liam’s unfortunate genetic mutation that resulted in a complete knockout of the Cohen syndrome protein would lead to a more severe developmental outcome. We were delighted to hear that it would not, as Dr. Wang has taken care of siblings with identical mutations who have had markedly different intellectual and physical capacities.

The Cohen Syndrome Association Conference in Cleveland has been rescheduled for 2021, and we plan to attend. We can’t wait to meet Dr. Wang in person, and to continue to learn from his research and experience.

– Ashley Waterman
Proud mother of Liam

Cohen Syndrome and DDC Clinic

DDC Clinic has helped to advance the understanding of Cohen syndrome in many ways. Through patient care, collaborations and scientific contributions, we have become recognized as a leading international medical facility for this rare disorder.

Patient Care

Our clinic has seen almost a hundred patients with Cohen syndrome. By serving these special children and families – Amish and non-Amish; from the United States, Canada, Europe, Australia and New Zealand – we have learned so much.

Community Support

In 2004, DDC Clinic and Dr. Wang organized the first Cohen syndrome gathering to share information with local families affected by this rare disorder. By 2008, with DDC Clinic’s help, a group of local parents started the Cohen Syndrome Association which has since evolved into a valuable resource and international presence. Working together, we present at CSA’s biennial conference and continue to provide hope, knowledge and support to families around the world.

Research and Collaborations

The scientific contributions we’ve made come from our international collaborations, not only with medical teams and research scientists, but most importantly, with children and families affected by Cohen syndrome.

Resources for Physicians and Families

We’ve shared our knowledge of Cohen syndrome through contributions and updates to online resources for the National Institutes of Health, NORD (National Organization for Rare Disorders), Orphanet, Human Disease Genes, Face2Gene, London Medical Databases Library, and GeneReviews.

Cohen Syndrome Association Conference Rescheduled

Due to the COVID-19 pandemic, the Cohen Syndrome Association (CSA) canceled its June 2020 conference, rescheduling the event for June 2021.

Held every two years in Cleveland, the CSA conference provides families affected by Cohen syndrome an opportunity to learn from medical professionals like Dr. Wang as well as many other international experts such as pediatric hematologist Dr. Larry Boxer, pediatric ophthalmologist Dr. Elias Traboulsi and pediatric orthopedic surgeon Dr. Dennis Weiner. Drawing attendees from all over the United States and around the world, the conference offers many families their only chance for an in-person appointment with Dr. Wang.

To assist families who would miss their appointment this year, DDC Clinic is offering telehealth visits with Dr. Wang. While telemedicine services are not new, most insurance companies in the past weren’t paying for those visits. Because of COVID-19, many insurers are now expanding their policies around telehealth services. If you’d like to schedule a telehealth appointment with Dr. Wang, please contact us at 440-632-1668.

If you would like to learn more about telehealth visits, call DDC Clinic at 440-632-1668.
What is Cohen Syndrome?

DDC Clinic has served Cohen syndrome patients for more than 20 years, and we’ve helped many families and physicians around the world. But what exactly is Cohen syndrome and what causes it?

Cohen syndrome is an inherited disorder affecting many parts of the body. Although signs and symptoms can vary widely, Cohen syndrome is characterized by developmental delay, intellectual disability, small head size and weak muscle tone.

Distinctive facial features include thick hair and eyebrows, long eyelashes, wave-shaped, down-slanting eyes, a bulbous nasal tip, a smooth or shortened area between the nose and upper lip, and prominent upper central teeth.

Worsening nearsightedness, degeneration of the light-sensitive tissue at the back of the eye, and an unusually large range of joint movement are common. Additional signs and symptoms may include low levels of white blood cells, overly friendly behavior, and obesity around the torso in late childhood or adolescence.

Frequency
The exact incidence of Cohen syndrome is unknown. It has been diagnosed in fewer than 1,000 people worldwide, and there are likely more cases which are undiagnosed.

Causes
Cohen syndrome is caused by mutations in the VPS13B gene (also called the COH1 gene) which are inherited in an autosomal recessive pattern. Recessive genetic disorders can affect an individual when both parents are carriers, meaning each parent carries a copy of the mutated gene. For Cohen syndrome to occur, each parent would have to pass their altered copy of the VPS13B gene to their child. As carriers, parents typically don’t show signs and symptoms of the disorder. If a child inherits one normal gene and one mutated gene, he or she will also be a carrier with no symptoms.

The Role of VPS13B Protein
Genes are involved in the production of proteins which are essential to the body’s many functions. The VPS13B protein is involved in glycosylation, which occurs when sugar molecules chemically attach to proteins and fats. This process forms glycoproteins and glycolipids which are important for healthy tissues and organs. The VPS13B protein is thought to be involved in normal growth and development of nerve cells and fat cells, and may play a role in the storage and distribution of fats in the body.

With Cohen syndrome, mutations in the VPS13B gene are believed to prevent the production of VPS13B protein and impair normal glycosylation. However, it is not known how a lack of VPS13B protein leads to signs and symptoms of Cohen syndrome. Researchers believe that problems with neuron development may be responsible for small head size, intellectual disability and retinal problems, and that abnormal fat storage may cause truncal obesity.

Treatment
Treatment for Cohen syndrome patients is highly individualized and tailored to an individual’s specific symptoms. Therapies and treatments can be complex and often involve a team of medical specialists. At DDC Clinic, we work with orthopedists, ophthalmologists and other healthcare professionals to achieve the best outcomes and help our Cohen syndrome patients reach their potential.

This article uses material from the NIH National Library of Medicine and NORD (National Organization of Rare Disorders). DDC Clinic and Dr. Wang helped to develop these online materials.

Scientific Contributions
Over the last 18 years, DDC Clinic’s research commitment to this disorder has led to four publications to better understand the condition and advance medicine:

Through the Eyes of a European Genetic Professional

By Helen Batchelor-Regan, PhD

Dr. Helen Batchelor-Regan is a trainee genetic counselor at the University of Manchester in England. She works at Birmingham Women’s Hospital in the largest clinical genetics department in the United Kingdom. She earned a PhD in Physiology from the University of Oxford before she fell in love with genetics. Helen traveled thousands of miles last fall to spend three weeks at DDC Clinic. We’re happy to share her perspective of our clinic.

First and foremost, I would like to thank DDC Clinic, other local healthcare providers who warmly hosted me and all the patients and families that greeted me so welcomingly whilst allowing me to sit in during clinic visits.

As a trainee genetic counsellor within the U.K.’s National Health Service (NHS), I’m encouraged to go outside of my training department to broaden my experience within the field. In the fall of 2019, I was lucky enough to experience and learn from the “DDC way” of providing healthcare to families affected by genetic conditions.

When I learned of DDC Clinic, I felt it would be the perfect place to achieve this. Not only does the clinic focus on providing genetic diagnoses to patients and their families, it also focuses on research to discover new genes that play a role in rare diseases, and international collaborations to develop understanding and treatments. I was amazed that a small clinic in a rural area could provide so much!

DDC Clinic’s set-up differs greatly from genetics services within the United Kingdom. Where the U.K.’s genetics services often refer patients to other departments for assessment and treatment of specific health concerns, DDC Clinic provides more holistic support to patients and their families. I was taken by how DDC Clinic aims to do so much in-house; offering primary care services, providing treatment options to alleviate symptoms and inviting specialists to run clinics from DDC Clinic itself.

This offers families the expertise needed close to home in a familiar environment. I felt this was key to supporting patients and their families by alleviating the need to build connections with many healthcare providers and having to explain their healthcare journey time and time again.

At DDC Clinic, families have a one-stop shop, with the team able to diagnose a simple ear infection whilst tweaking medications to ease suffering all in one visit. This allows the team to understand these conditions better than most other healthcare professionals, enabling bespoke care and treatments to be offered.

My experiences at DDC Clinic far surpassed my expectations. I was struck by the welcoming feel of the clinic – a place where all who came could be at home. The connection between DDC Clinic’s team and families was invaluable. The trust and warmth in these relationships only goes to demonstrate the value of the clinic and its ethos. It was clear how valued the clinic (built by the community, for that community, and the benefit of others) is to the community itself.

During my visit I was able to see the benefits of the Amish Genetic Disease Panel for testing, encompassing the main causes of genetic conditions within the Amish communities. This enables testing to give a rapid answer to families, preventing the pain of a long, drawn out diagnostic journey.

The hard work and research at the clinic has enabled the development of this bespoke test, and the further efforts towards understanding these conditions to improve treatment helps to ease suffering. I feel that this is the way forward for genomic medicine. A journey does not end with the diagnosis, and therefore DDC Clinic’s whole person approach is crucial for improved treatment. I hope to maintain my relationship with DDC Clinic for years to come.
Early this year, our molecular diagnostics laboratory expanded our Amish Genetic Disease Panel, enabling us to simultaneously test for 160 rare conditions found in the Amish and Mennonite populations.

The expanded panel was completed months ahead of schedule, thanks to Dr. Bao Xin, our Research and Technical Director who led the project, and DDC Clinic’s Valerie Sency, Karen Cechner and Julia Szekely.

“We’re very excited to be able to now screen for an additional 50 conditions as part of this panel,” says Dr. Wang. “Early diagnosis leads to early treatments, and those effective treatments can be lifesaving.”

In partnership with the Care Center in Middlefield, the panel is also being used for DNA-based newborn screenings. Using a sample of a newborn’s cord blood, our lab can determine if a baby is affected by any of the 160 rare conditions. If a condition is identified, no further testing is needed and support services can begin. Thanks to our donors and the United Way of Geauga County, parents pay only $25 for testing as part of their birthing fee.

The Elisabeth Severance Prentiss Foundation and the Fowler Family Foundation partially funded the project, making it possible to keep the cost of the new panel the same as the previous one.

The recently passed stimulus legislation made changes which can benefit individuals, corporations, and charities including DDC Clinic. For charitable contributions made in 2020, the highlighted changes include:

- Taxpayers who take the standard deduction on their 2020 tax return can claim a $300 ($600 for married filing jointly) deduction for cash gifts made to certain types of charitable organizations which include DDC Clinic.
- For 2020, individuals may deduct charitable cash donations up to 100% of their adjusted gross income (if made to certain types of charitable organizations).
- Certain types of corporations may deduct charitable cash donations up to 25% of taxable income.
- Required minimum distributions (RMD) for individual retirement accounts (IRAs) and most defined contribution plans are waived for 2020. If you are at least 70 ½ years old, you can still make tax-free charitable donations directly from your IRA. You can benefit DDC Clinic while excluding up to $100,000 annually from gross income by making a qualified charitable distribution (QCD).

DDC Clinic does not provide tax or legal advice. Please consult your tax preparer and/or attorney for additional guidance.

Amish Genetic Disease Panel Tests for 160 Conditions

Welcome Back, Carol

We’re pleased to welcome back Carol Troyer Counselman as our new part-time RN.

Fluent in Pennsylvania Dutch, Carol began working at DDC Clinic in 2006 writing our Amish newsletter. After getting certified as a medical assistant and phlebotomist, Carol left in 2010 to work in a hospital emergency department and then earned her nursing degree from Kent State University in 2016.

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Carol Troyer Counselman

“Working in a fast-paced environment for nine years in the E.D., with nearly three of those years as a nurse, has provided me with valuable experience and taught me a lot, not only about life and death, but also how intricately we’re created by God,” says Carol. “I’m happy to be back at DDC Clinic and hope to apply my knowledge and skills as a nurse by caring for patients and assisting with ongoing research.”

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- Linda Vinney
- Joe Vitale
- Crist Yoder
- Robert Yoder

## By the Numbers

- **16 Years**
  - Providing family support meetings
- **17 Years**
  - Providing care for children with rare disorders for over 17 years
- **120 Minutes**
  - Initial office visit to help families find answers
- **Nearly 1,100 Patients**
  - Served to date
- **4,400 Children**
  - Immunized over 10 years
- **55**
  - Patient centered research publications
- **863 Disorders**
  - Genetic testing for
- **2.5 Million Dollars**
  - In free and reduced cost prescriptions for families through our patient assistance program in the last year
- **750 Patients**
  - Served annually in specialty clinics including Neurology, Dental and Hemophilia

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