

Finding Answers Through Collaborative Research

Most of you know DDC Clinic for the comprehensive and highly personalized medical care that we provide to our patients. However, many of you may be surprised to learn that for the past several years our clinic has been partnering with one of the world's largest genetic research centers on an important project that has significantly impacted the lives of many of our patients.

In 2016, Regeneron Genetics Center (RGC), a subsidiary of Regeneron Pharmaceuticals, Inc., invited DDC Clinic to participate in a multi-year, large-scale research project involving whole exome sequencing (over 20,000 genes in a human genome) of DNA samples to determine the genetic factors that cause or influence a wide range of human diseases.

Specifically, the DDC-RGC partnership involves working together to study undiagnosed rare conditions in the Amish population. Since the project began, previously unknown gene mutations have been identified, enabling our clinic to uncover the underlying genetic causes of rare diseases affecting some members of our community.

"This partnership has proven to be very beneficial to us and to the patient families we serve," says Dr. Wang, DDC Clinic Medical Director. "It has produced new discoveries that have resulted in life-changing outcomes, not only for our patients, but also for others around the world."

As an integral component of the research project, DDC collects patient-consented DNA samples from our Amish patients and their family members. RGC conducts state-of-the-art whole exome gene sequencing on the samples free of charge and sends back the raw data to our molecular diagnostics laboratory for further analysis.

"Integrating Regeneron's whole exome gene sequencing into our diagnostic process has allowed our in-house lab to leverage highly advanced genomic tools to find answers to our most difficult cases," says Dr. Wang. "Many of our patients were undiagnosed for years and faced significant challenges. Because we've been able to work with this extremely important



Dr. Baozhong Xin, Research & Technical Director, DDC Clinic

laboratory data, our clinic has been able to provide many of our patient families with long-awaited answers and a path toward understanding."

To date, DDC Clinic has received genomic data for more than 1,100 individuals, including 158 patients with undiagnosed conditions. We've diagnosed 46 previously unknown disease genes and decisively diagnosed 66 patients. In addition, we've identified potential candidate genes for 36 patients. Those numbers will grow as the project continues and more discoveries are made.

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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 1,500 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.

My Journey with DDC Clinic

After being part of DDC Clinic for 16 years as a board member, I was thrilled, yet humbled, to be offered the position of Executive Director. I certainly had no way of knowing, back in 2003, when my family's journey began with DDC, that it would become such a huge part of our lives.

I remember hearing about Das Deutsch Center (DDC) around 2000-2001. Of course, I paid little attention as my wife and I were newly married with our first precious little daughter. This new endeavor didn't concern us. This "Center" was for families with special needs children, and that didn't include us. We would never need it, so why should we be interested.

That all changed in 2002.

That year my nephew was diagnosed with Prolidase Deficiency, an enzyme disorder that causes painful skin ulcers and recurrent infections throughout the body. After much suffering and numerous hospitalizations, he passed away in January 2003.

Two weeks after he died, we received some alarming test results at a wellness check-up for our second daughter, born in August 2002. She ended up in the hospital, and after a few visits, she also was diagnosed with Prolidase Deficiency. That was the beginning of our journey with the DDC family.

In February 2005, my daughter underwent a stem cell transplant at Rainbow Babies and Children's Hospital. She was the first patient with Prolidase Deficiency to have the transplant, and the doctors there felt that there was a very good chance that she would be cured.

It was during that time that I was invited to a Board meeting at DDC Clinic, becoming a board member in June 2005. Looking back, I don't remember a lot of the details about how our journey brought the clinic to where it is today. What I do remember is the support DDC received from our community, our family and friends, and various foundations and organizations. I also remember the many willing hands that helped build our new clinic, raise funds and put on our benefit auctions. The list goes on and on.

A simple thank you to all who helped over the years seems very inadequate, but we mean it with our whole hearts.

My daughter's stem cell transplant was not a cure for Prolidase Deficiency, and so far, no cure has been identified. Is that why I took this role of Executive Director? To try and prod our doctors to work harder to find an answer? Certainly not.

Our Research Leads to New Discoveries

By leveraging today's most advanced genomic tools, DDC's molecular diagnostics laboratory has been able to provide answers to our patients affected by previously undiagnosed conditions. We anticipate making more progress as our ongoing collaboration with Regeneron Genetics Center continues. Genes and disorders that we have studied and identified through the collaboration include:

Gene	Disorder
SCAPER	Intellectual developmental disorder and retinitis pigmentosa
MECP2	Rett syndrome
POGZ	White-Sutton syndrome
ADNP	Helsmoortel-Van der Aa syndrome
NSD1	Sotos syndrome
PTEN	Cowden syndrome
GNAO1	Developmental and epileptic encephalopathy-17
TANC2	Intellectual developmental disorder with autistic features and language delay
ZBTB18	Autosomal dominant mental retardation-22
CNNM4	Jalili syndrome
CHD2	Developmental and epileptic encephalopathy-94
DSP	Dilated cardiomyopathy
NT5C2	Autosomal recessive spastic paraplegia-45
NIPBL	Cornelia de Lange syndrome
SYNGAP1	Autosomal dominant mental retardation-5
HNRNPH2	Bain type of X-linked syndromic intellectual developmental disorder
PNPT1	Combined oxidative phosphorylation deficiency-13
HYDIN	Primary ciliary dyskinesia-5

Gene	Disorder
FOXP3	X-linked immunodysregulation, polyendocrinopathy, and enteropathy
SLC13A5	Developmental and epileptic encephalopathy-25 with amelogenesis imperfecta
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures
KMT2C	Kleefstra syndrome-2
TBR1	Intellectual developmental disorder with autism and speech delay
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency
ZIC2	Holoprosencephaly 5
KMT2D	Kleefstra syndrome-2
PURA	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties
PI4KA	Neurological, intestinal and immunological disorder
HNRNPU	Developmental and epileptic encephalopathy-54
TMEM38B	Osteogenesis imperfecta type 14
ZMYND11	Autosomal dominant mental retardation-30

My Journey with DDC Clinic > from page 2

We realize that answers may never be found for our children. But maybe one day your children, grandchildren, or great grandchildren will be born with a devastating disorder and DDC Clinic will have found a treatment or even a cure.

DDC Clinic is not about treating one certain disorder. It is so much more. It's a place where our community can gather to love, share and care about one another; a place of compassion, hope and faith.

My vision for the future of DDC Clinic is to have our doctors be the first thought that comes to mind when parents suspect that something is not as it should be with their child. When parents worry that their child is not okay, maybe a simple blood test can reassure them that he or she is healthy. If their child is sick, we

can provide the care and resources parents need so, if possible, their child does not need to be hospitalized.

I look forward to working with all of you and the whole DDC community, and to continuing our important mission. Please remember DDC Clinic in your prayers.

Eli Miller



Executive Director
DDC Clinic – Center for Special Needs Children



Juliz Szekely, Senior Laboratory Technologist



Closing the GAP in Genetic Awareness

DDC Clinic's molecular diagnostic laboratory has officially changed the name of our Amish Genetic Disease Panel to Genetic Awareness Panel, or GAP.

"We felt it was important to change the name to Genetic Awareness Panel to make it more meaningful to our community and to move the focus away from disease and more on knowledge and awareness," says Dr. Heng Wang, DDC Clinic Medical Director.

"We hope that creating greater awareness will lead to more early diagnoses. All too often, investigating genetic disorders in our Amish patients begins when a patient's disorder has already progressed significantly," continues Dr. Wang. "But with early diagnosis, we can have earlier intervention and treatments, improved health outcomes and less burdensome medical costs."

Even though the name has changed, GAP still remains the same groundbreaking diagnostic tool that can simultaneously test for more than 160 rare genetic conditions found in the Amish population. GAP offers accurate and affordable testing for both children and adults. When performed as a supplement to state newborn screenings, GAP can detect and provide early diagnosis of many conditions, allowing infants to begin specific treatment sooner. For Amish adults, GAP testing provides awareness of an individual's predisposition to genetic disorders and identifies if they're carriers for diseases which could get passed on to their children.

Since its start, GAP has proven to be an important tool, both for diagnosis and awareness of risk factors. To date, our clinic's molecular diagnostics laboratory has used GAP to provide answers to patients throughout the Midwest affected by a variety of conditions including GM3 Synthase Deficiency, Byler Disease, Cohen Syndrome and Prolidase Deficiency. We've also identified children who are at increased risk for developing a blood clot, heart condition or other issue as an adult, information that is critically important to parents, providing knowledge of what their children may experience in the future.

GAP is a great example of personalized medicine in action. We're so excited to have this crucial tool, as we're not only closing the gap in genetic awareness, but also the gap between patients and physicians; the plain community and the healthcare system; rare genetic disorders and common diseases; disease diagnosis and treatment and disease prevention.

If you're interested in GAP testing but you're not a DDC patient, you'll need to work through your family physician. For more information on the benefits of GAP, contact DDC Clinic at 440-632-1668.

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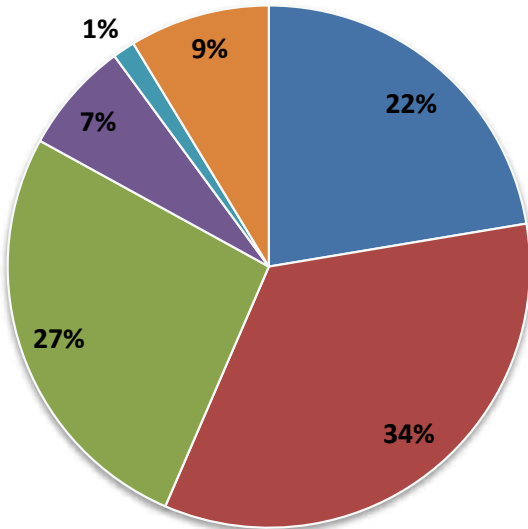
Our partnership with RGC is only one aspect of our work. Many of our patients, supporters and friends may not know that our modest clinic has been recognized internationally for our state-of-the-art genetic research impacting broad-based health concerns such as heart disease, developmental disabilities, and neurological disorders. Collaborating with more than 15 major institutions around the country and Europe, we've made tremendous progress in identifying many previously unknown genetic diseases. Since opening our doors in 2002, over 200 genetic diseases have been identified in our community, producing life-changing results.

Our clinic is small but mighty, and we continue to grow. Our ongoing clinical research and collaborative partnerships are vital, enabling us to improve diagnoses of rare conditions and provide better care for the special needs children and their families who need our help.

2020 FINANCIAL REPORT

Revenue

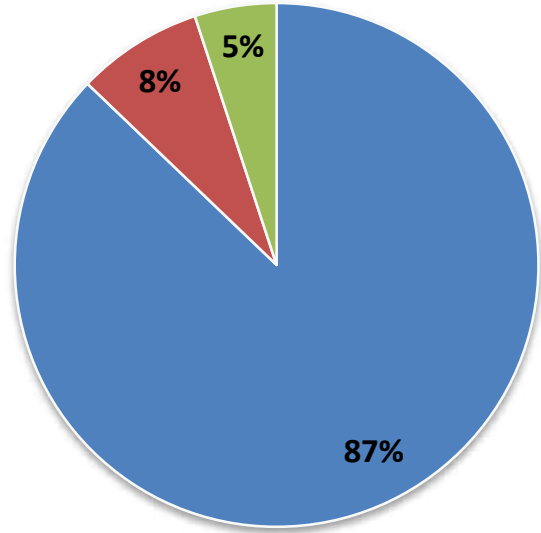
Total Revenue for 2020 were \$1,386,298



■ Contributions	\$310,086
■ Fundraisers	\$473,052
■ Clinical Services	\$367,437
■ Research Contracts	\$96,077
■ Miscellaneous Income	\$19,217
■ Individual Giving	\$120,429

Expenses

Total Expenses for 2020 were \$1,193,313



■ Program Services	\$1,040,082
■ Fundraisers	\$92,951
■ Management & General	\$60,280

Greg and Scotty's Story

This is our family's story about my son, Greg, and my grandson, Scotty, and how they both battled a severe illness that went undiagnosed for many years, until we finally got the answer we had been searching for.

Our story starts when our son Greg, little brother to our firstborn, Lauren, was born in April 1989 in Missouri. He was a robust 9 lb. boy at birth. He was colicky the first few weeks and had frequent loose stools, but our pediatrician wasn't concerned as our little boy was gaining weight. At 8 weeks of age, he began to experience vomiting, lethargy and weight loss in addition to diarrhea. Greg was admitted to a children's hospital in St. Louis.

Routine testing was negative for obvious infections and parasites, but our young son continued to decline. Doctors identified his condition as "intractable diarrhea of infancy," a condition most common in third world countries. Greg was taken off all oral feedings and placed on TPN (Total Parenteral Nutrition) so he could intravenously receive the nutrition his body needed.

On top of everything that was going on during that terrible time, it seemed that hospital staff and other family members suspected or blamed us for Greg's elusive condition. We were emotionally distraught and frustrated by the lack of answers, and we were painfully insulted that anyone would accuse us of harming our own precious child.

During the two and a half months that our little Greg was hospitalized, testing detected an erosion of villi in his small intestine, but the cause remained unknown. He was slowly returned to oral feedings of pre-digested formula, first by a feeding tube through the nose, then by mouth. When Greg was finally discharged from the hospital, his doctors warned us that he would be very vulnerable to gastrointestinal infections, and

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he shouldn't be in day care. I made the decision to leave my job to care for Greg at home.

In early childhood, Greg also suffered from multiple allergies, eczema and asthma. His pediatrician labeled it a possible "autoimmune disorder." Ultimately, Greg outgrew many of his problems, and we feel very blessed that he is now a healthy adult male.

Enter Scotty

Fast forward to December 2016. Our daughter Lauren, who had relocated to the Cleveland area, gave birth to her second child, Scotty. She too had a healthy daughter as her firstborn. Scotty was tiny at birth, only 5 lb, 11 oz. He had noticeably loose and frequent stools and initial weight gain was slow.

At about 6 weeks of age, Scotty developed a skin infection, and he was admitted to the hospital for treatment. The weeks that followed were a blur of hospitalizations. Scotty couldn't gain weight and was labeled "failure to thrive." He was repeatedly dehydrated despite constant efforts at feeding. His skin was dry and scaly, and he was diagnosed with possible ichthyosis, a genetic skin disorder. He also suffered from severe metabolic instabilities.

After some time, blood testing revealed Scotty had aldosterone synthase deficiency. Ultimately, a gastronomy tube was surgically placed to ensure he would get the nutrition he needed, and this feeding tube remained in place over two years.

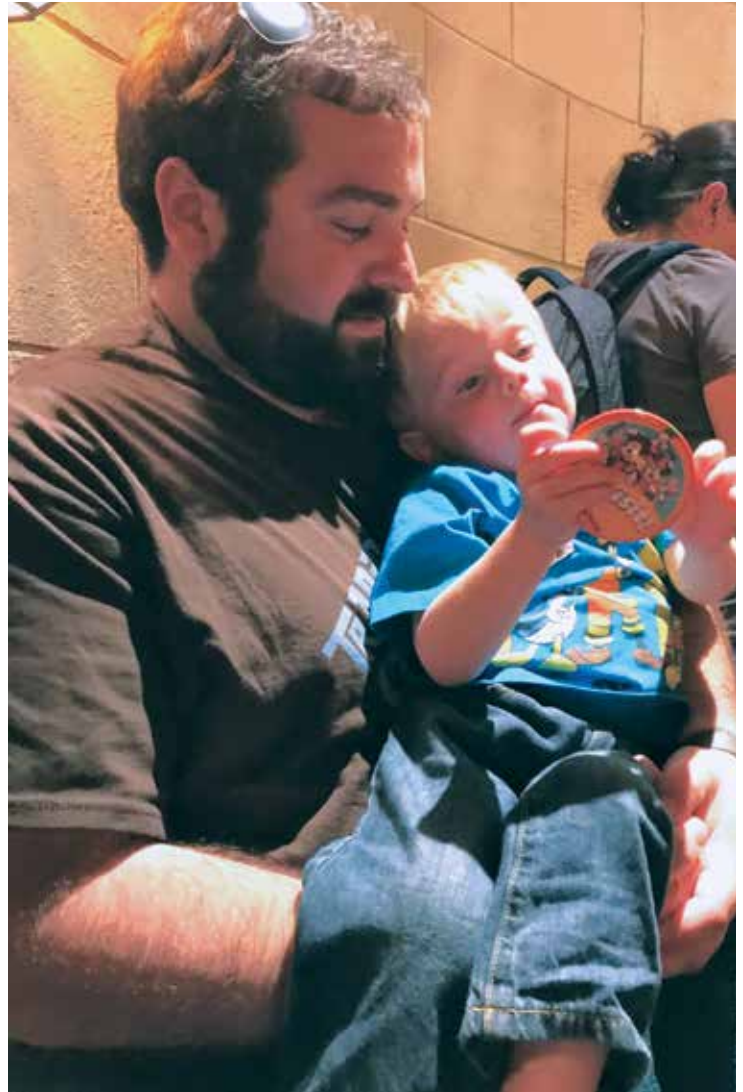
As Scotty's maternal grandparents, my husband and I made frequent trips to Cleveland to help with Scotty's care, and to give respite to our daughter, her husband, and Scotty's paternal grandparents, Scott and JoAnn Brace, who were also helping with Scotty.

I couldn't help but see that Scotty's condition drew many familiar parallels to my experience with our son, Greg, and all the same feelings of frustration and helplessness came flooding back. I spoke to physicians about the similarities between Scotty and his Uncle Greg, but basic genetic testing didn't detect any specific abnormalities.

Long-Awaited Answers

One day, Scotty's grandparents, JoAnn and Scott, who have been active on DDC's Board, told Dr. Wang about Scotty and Greg. He felt the parallels couldn't just be coincidental. He invited us to come to the clinic and listened closely to our stories.

Dr. Wang expertly led the effort to unlock the medical mystery that had so significantly affected our family. He enrolled Scotty



and Greg in a detailed genetic study with Regeneron Genetics Center. The answer we had been searching for came when Scotty was nearly 3 years old. Greg and Scotty were both diagnosed with a form of IPEX Syndrome: Immune Dysregulation, Polyendocrinopathy, Enteropathy, X-Linked.

It was such a relief for us to finally "name it." We now understand why these two generations of male children suffered life-threatening conditions in infancy. We also know for future generations of our family how to better diagnose and care for children who may suffer from the symptoms of this disorder. We're encouraged that both Greg and Scotty have attained improved health with age. We're grateful to DDC Clinic and Dr. Wang for providing the answers our family needed.

[Susan M. Buerkle, Scotty's maternal grandmother](#)

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