DDC clinic
Center for Special Needs Children

Spring 2022
Chronicle

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

Cardiomyopathy – One Family's Story

Caleb's Journey

"Your test is positive" were the welcome words we heard when we tested for pregnancy. We were still grieving the loss of our stillborn child several months prior, so with this good news came anxiety and the question, "Is everything okay?"

Months went by with doctor visits and ultrasounds. A stress test graph showed that our baby's heart was not responding to stress as it should. After an overnight hospital stay to monitor the baby's heartbeat, we were told "everything is fine."

Six days prior to full term, our doctor noticed a low fluid level and decided to induce labor. The emotions we felt after our baby's first cry were indescribable to say the least. After all the anxiety and tests, we thought we had a healthy baby. We named him Caleb.

Three days later, a visiting nurse noticed Caleb's fast breathing and called our family doctor, who thought it was okay to wait a week, until our scheduled appointment, to check it out.

But on the tenth day of Caleb's life, his chest started retracting and his breathing increased to one hundred times per minute. We took him to the local hospital where an x-ray showed an enlarged heart. He was transferred to Akron Children's Hospital, where they performed minor surgery to insert IVs, totally paralyzed him, and put him on a breathing machine.

After many tests, we received a diagnosis – hypertrophic cardiomyopathy, for which there is no cure. We were told that Caleb would not live longer than a year. There was nothing that could be done.

We also found out that he had restrictive cardiomyopathy and possibly metabolic cardiomyopathy. The fibers in his heart muscle were not lined up properly causing them to work in different directions, resulting in a weak heartbeat and enlarged heart. It also caused his lungs to not generate enough oxygen, creating fluid buildup behind his lungs – the adult equivalent of congestive heart failure.

It truly was a medical marvel that Caleb was alive at the end of that first day in the hospital. We were thankful for the doctors and nurses who used their God-given abilities to maintain life. On the sixth day of his hospital stay, we were told that Caleb may not make it through the night. That night we committed him and released him to God.

The next morning, Caleb was still alive! It was an Abraham and Isaac experience, reminding us of how Abraham must have felt when God said that he didn't need to offer up his son after all. Caleb started to improve, and we took him home.

Around this time, we learned about a genetic doctor in Geauga County who had success with treating heart patients nutritionally. Here was hope! Coming from a big city hospital to the humble DDC Clinic convinced us that it was one of the best kept secrets in Ohio. Our initial visit, which lasted several hours, was encouraging. Dr. Wang listened not only to our medical needs, but also to our emotional and spiritual concerns.

We treasured every day we had with Caleb, making him as comfortable as possible, and giving feedings and medications every three hours. After a month at home, Caleb quietly passed away in the wee hours in his mother's arms. He was seven weeks and three days old.

While Caleb did not respond as well to treatment as some other children, we feel blessed by our experience with DDC Clinic. We knew Caleb had a purpose, no matter how short his life was. God knows who our parents need to be to form us to fulfill the purpose He has for each one of us.

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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 1,500 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.

Executive Director's Corner

Reflections

Taking a look back at the first 20 years of DDC Clinic's existence, I must marvel at what has been accomplished.

I wonder, when the founders held the first meeting to discuss what they envision for such a clinic, did they think about what they hoped to accomplish in 20 years? Did they ever feel like "this is never going to happen?" They certainly couldn't know what the future would hold, but they had a vision, and a lot of faith.

And they also had to know that all good things take time, and most likely younger generations would benefit more from their endeavor than they themselves would. I have asked myself many times, am I thankful for the sacrifices they made? Do I appreciate their unselfish work? The persistence and dedication that made this possible?

So, what was the recipe for success at DDC Clinic for the past 20 years? I'm hardly qualified to answer that question, but in my mind it was the ever-present faith, hope, love, compassion and teamwork that we constantly witnessed, and above all, the Grace of God. This just doesn't happen without God's blessing. And I truly believe if we continue to follow and practice these, we will continue to receive His blessings, and He will allow us to continue this work through future generations.

As we head into summer in Northeast Ohio, we are also entering our benefit and auction season. It is so humbling and rewarding to see all of you show up at these events. Please see our events list in this newsletter and mark your calendars.

Thank you to our board, staff, funders, community, volunteers, donors and supporters. You are all greatly needed and appreciated. Each of you has been a blessing to DDC Clinic and the special children, and people, we serve.

Eli Miller
Executive Director
DDC Clinic

Genetic Awareness Panel Helps Our Community

My name is Carol Counselman, and I'm a nurse at DDC Clinic. I'm also a member of the Anabaptist community, which helps me in my interactions with our patients. I wanted to tell you about my work involving our Genetic Awareness Panel and how important the panel is to our community.

As you may know, DDC Clinic changed the name of its genetic panel test from Amish Genetic Disease Panel to Genetic Awareness Panel, or GAP for short. This was done, in part, to help clarify that this test is available to the Anabaptist community as a whole, not only Amish but also others such as Mennonites.

The development of the Genetic Awareness Panel (GAP) began nearly 20 years ago through the Love, Faith, and Family Project supported by the Robert Wood Johnson Foundation. The primary objectives of this initiative included raising awareness of genetic disease and risk in the Amish community as well as establishing early diagnosis and treatment of newborns with genetic disease.

After a survey was conducted among members of the Amish community, it was determined that one of the greatest needs was to disseminate information regarding inherited disorders, as well as an increased interest among families for genetic carrier testing. From this project, GAP was born and developed in 2018, after which DDC Clinic initiated a pilot program in 2019 at the local Amish birthing center known as the Middlefield Care Center (MCC).

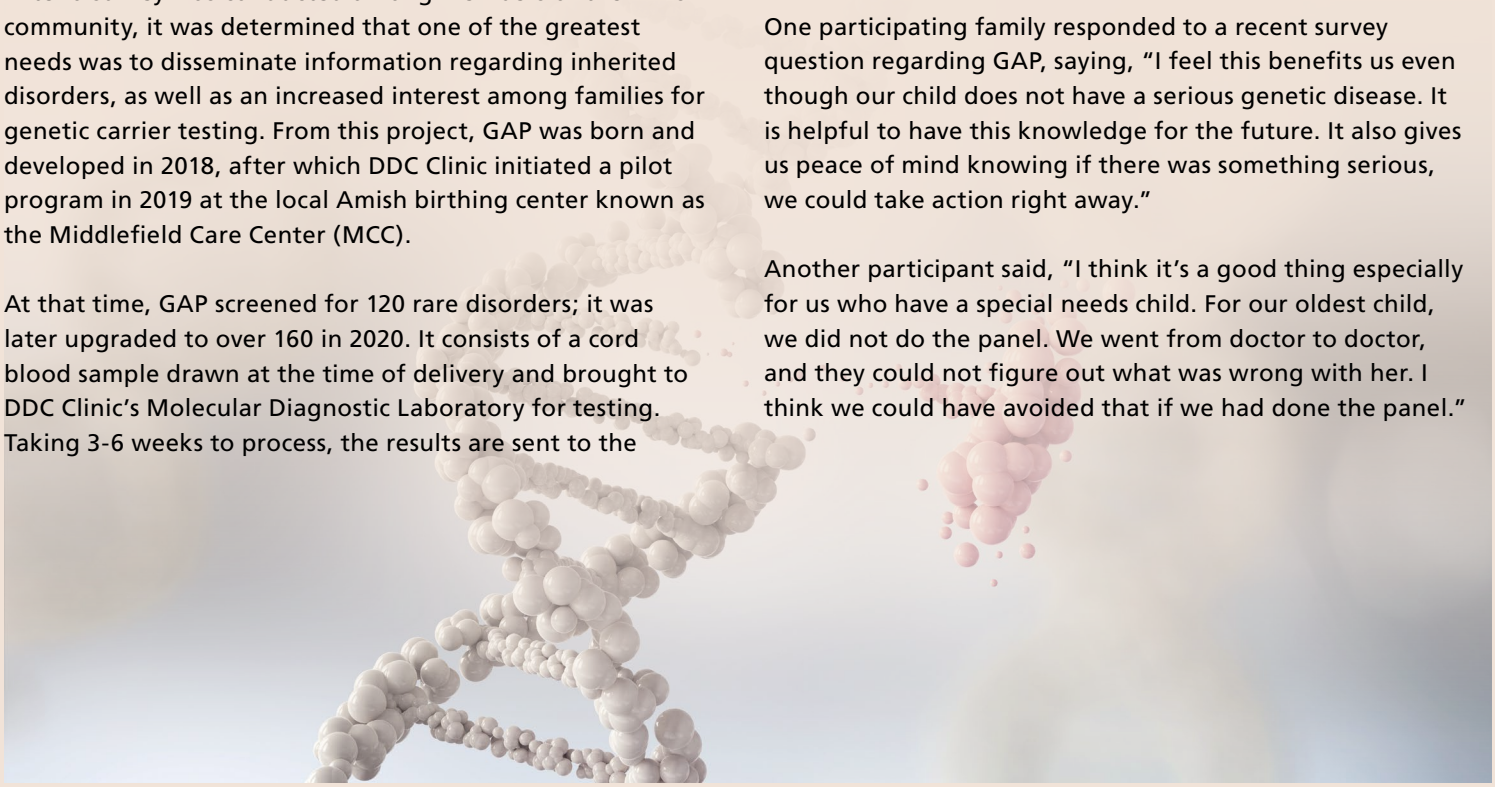
At that time, GAP screened for 120 rare disorders; it was later upgraded to over 160 in 2020. It consists of a cord blood sample drawn at the time of delivery and brought to DDC Clinic's Molecular Diagnostic Laboratory for testing. Taking 3-6 weeks to process, the results are sent to the baby's pediatrician, who reports the findings to the family. This establishes awareness of any impending, immediate concerns that may be associated with genetic test results. It also allows for early diagnosis and treatment; reduces unnecessary hospital admissions, testing, and medical costs; and helps minimize undue worry and anxiety.

To help explain the positive impact that GAP can have on our community, I personally meet with expectant Amish mothers during their prenatal appointments at MCC. Speaking Pennsylvania Dutch, I provide helpful information on how parents can utilize this supplemental newborn screening at the time of their baby's birth.

This often leads to further questions and discussions regarding family history involving such disorders as hypertrophic cardiomyopathy, factor V Leiden, hemochromatosis and long QT syndrome. With over a 90% participation rate in this service offered at MCC, families are able to screen whether their newborn is affected by a rare genetic disorder or only a carrier.

One participating family responded to a recent survey question regarding GAP, saying, "I feel this benefits us even though our child does not have a serious genetic disease. It is helpful to have this knowledge for the future. It also gives us peace of mind knowing if there was something serious, we could take action right away."

Another participant said, "I think it's a good thing especially for us who have a special needs child. For our oldest child, we did not do the panel. We went from doctor to doctor, and they could not figure out what was wrong with her. I think we could have avoided that if we had done the panel."



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A New Life

Time passed, and we were expecting another child. We were concerned that this child could also be affected by hypertrophic cardiomyopathy. We had two options for testing: Taking amniotic fluid before birth or collecting cord blood at birth.

We opted for using cord blood. No matter what the outcome, testing was a win-win. If the test was negative, we would know right away that our child was healthy. If it was positive, we could keep our child comfortable at home, eliminating the need for a hospital stay.

Our daughter was born by C-section and within minutes started turning blue and having trouble getting oxygen. We just "knew" that we were going down the same road as before.

I planned to ship our baby's blood sample "Next Day Delivery" to DDC Clinic since we were two hours away. When the nurse asked about our preferences on transferring our baby to Akron Children's Hospital, I had to have her blood tested NOW! A taxi driver was summoned, and DDC Clinic had the sample that day.

With the supreme effort that the DDC Clinic team put in place, they were able to get the results in an amazingly short period of time. "She'll be fine!" were the best words we ever heard Dr. Wang say.

The value of genetic research and testing cannot be overstated. Now anyone can have a blood sample drawn to see if they carry this disease. In our case, we were able to go home from the hospital knowing that if our child got sick, it wasn't because of hypertrophic cardiomyopathy.

While it is hard to talk about the financial end of things, as it seems irrelevant when it involves "quality of life" for special children, the fact remains that the sooner you diagnose and treat your child, the more savings you have for the long term health care costs of our community.

We thank everyone at DDC Clinic for the passion they have in fulfilling their mission. May God bless!

~Myron and Ruth Troyer

It's not too late to purchase your tickets to our annual Patchwork Benefit on Saturday, June 4 at Meadow Ridge Events in Windsor, Ohio.

Join us for a special evening commemorating our *20-year journey of hope, faith and caring.* Enjoy food, drinks, music, auctions and a special presentation about our work and the families we've helped.

Your support enables us to provide life-changing diagnoses and treatments to children affected by rare genetic disorders.

For tickets and more information, call 440-632-1668 or visit ddcclinic.org.

Calling All Low Protein Cooks

The La Farge Medical Clinic's Center for Special Children in Wisconsin is putting together a family-style low protein cookbook for the Plain community. Many families prepare low protein foods for loved ones with phenylketonuria (PKU), maple syrup urine disease (MSUD), propionic acidemia (PA), and glutaric acidemia type 1 (GA1).

If you prepare low protein foods for your family, the clinic is asking for tasty recipes from your kitchen to share with other families from the Plain community. This cookbook will be a valuable resource for all families who cook low protein meals – whether you've been cooking low protein foods for years or just getting started.

All types of recipes are welcome, including snacks, soups, main dishes, breakfast items, breads, noodles, desserts, kid-friendly foods and more. No recipe is too simple or too complex.

All recipes will be analyzed for leucine, phenylalanine and protein, and nutritional information will be included with each recipe in the book.

If you'd like to share a recipe, visit DDC Clinic to pick up a Recipe Submission Form or **call 440-632-1668**. There's no limit to the number of recipes you can submit. You can also request a Tip Form to share quotes, tips or other words of encouragement to be included in the book.

Recipe Submissions and Tip Forms must be sent to the La Farge Medical Clinic's Center for Special Children by June 30, 2022.

This project is funded by a grant from the Midwest Genetics Network, a program dedicated to improving access to genetics services. Once completed and printed, the low protein cookbooks will be available at no cost to Plain families who prepare low protein foods.

Cardiomyopathy Family Gatherings

DDC Clinic will host a family gathering on Saturday, July 23 for Amish families from Ohio and surrounding states who have lost children or have had a child with a heart transplant due to MYBPC3 gene-related cardiomyopathy.

This one-day gathering offers an opportunity for families to share, learn and support one another. Families will learn about the genetics of cardiomyopathy and get updates on current treatment options, ongoing research and potential future therapies, including gene therapy.

The program will feature discussions, Q&A sessions and presentations by DDC Clinic doctors and staff who will be joined by geneticists and pediatric cardiologists from other major medical centers such as Cleveland Clinic, University Hospitals and University of Pittsburgh Medical Center as well as representatives from Tenaya Therapeutics.

Families who attend will be offered free targeted *MYBPC3* carrier testing performed by DDC Clinic Molecular Diagnostics Laboratory.

The event will run from 8:30 a.m. - 5 p.m. Breakfast, lunch and childcare services will be provided at no cost. DDC Clinic will also help families with transportation costs as needed. Registration is required.

Look for more information coming soon to your mailbox or contact DDC Clinic at 440-632-1668 to learn more.

Together, We Can Change Children's Lives

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

Isaac's family is just one of nearly 1,500 families with special needs children that we've helped over the years.

Individually, each condition that our clinic identifies, diagnoses and treats is indeed rare; but collectively, rare genetic disorders are not. One in 10 of us will have a family member affected by a rare genetic disorder sometime during our lives.

Rare genetic disorders are our disorders and working together, we can make a big impact.

Thank you for your continued support. If you'd like to make donation, please use the enclosed envelope, visit ddcclinic.org or call 440-632-1668.

Understanding Genetics

Studying Pediatric Cardiomyopathy

Pediatric cardiomyopathy affects infants and children in our community and around the world, regardless of age, gender or ethnicity.

Cardiomyopathy is a rare heart condition in which the heart muscle becomes enlarged, thickened or stiffened, hampering the heart's ability to pump blood. Hypertrophic cardiomyopathy (HCM) is one form which affects 40-50% of children with cardiomyopathy. HCM, which often occurs in more than one family member and across generations, is often caused by mutations in the *MYBPC3* gene.

Genes are segments of DNA that provide instructions to create the proteins our body needs to function. Certain genetic mutations, such as those affecting the *MYBPC3* gene, disrupt these instructions and impact proteins that control the ability of the heart to pump blood through the body.

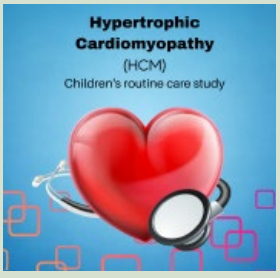
While cardiomyopathy has been diagnosed and understood as a condition for more than 100 years, over the past 20-30 years we've achieved a greater understanding of the genetic causes of the disease, thanks to clinical research, including natural history studies.

What are Natural History Studies?

In general, clinical research studies are designed to add to medical knowledge related to the treatment, diagnosis and prevention of diseases.

Natural history studies are an important way to gain deeper understanding as they track the course of a disease over time, identifying demographic, genetic, environmental and other factors that may affect how severe the disease becomes. A thorough understanding of what patients with a disease experience is the foundation of the development of new therapies.

Today, because of knowledge gained through research, we understand more about the role of genetics in cardiomyopathy than ever before and are closer to developing new therapies than ever before. However, the ability to develop new therapies is only as good as the information we have about the condition.



DDC Clinic's Contribution to Cardiomyopathy Research

One important way that our clinic gains greater understanding of conditions such as cardiomyopathy is through research partnerships with major medical institutions, research centers and biotechnology companies.

As such, DDC Clinic was the first to be chosen to participate in an important natural history study sponsored by Tenaya Therapeutics to help better understand the impact of *MYBPC3* genetic mutations on heart disease, including hypertrophic cardiomyopathy, in children.

The five-year study, conducted in 40 sites worldwide, is designed to collect and assess the medical data of patients with the *MYBPC3* genetic mutation. The data will enable researchers to characterize and evaluate disease course, burden of illness, risk factors and biomarkers associated with disease progression.

This study is an important step on the path to learning more about cardiomyopathy and answering the many questions families have about the condition, one of the most common causes of sudden death in adults and children.

"We're very pleased to be a part of this important global study on pediatric cardiomyopathy," says Dr. Wang, DDC Clinic Medical Director. "Contributing our patient data to this study will not only benefit our local families, but families around the world who have children affected by cardiomyopathy. The knowledge that we gain from this natural history study may one day provide much needed hope to parents and make a difference in the lives of our young patients."

The results of the natural history study are intended to inform the development and clinical trial design of investigational drug products, including gene therapies. In addition, the combined data and analysis from participating centers involved in the study may lead to scientific publications that ultimately contribute to the overall collection of information available on cardiomyopathy.

To learn more about hypertrophic cardiomyopathy, read one family's personal story on the front cover of this newsletter.

COVID Vaccine – Trust the Facts

There's a lot of false or misleading information that has spread about the COVID-19 vaccine, and you may have heard some of it. Be sure you know the facts so you and your family can stay safe. This helpful information, which comes from the Ohio Department of Health, separates the fiction from the facts.

FICTION	FACT
COVID-19 vaccines aren't safe.	COVID-19 vaccines are safe and effective.
COVID-19 vaccines were rushed and developed too quickly.	COVID-19 vaccine development and clinical trials were thorough, and thanks to a strategic scientific effort to streamline processes, developed more efficiently.
Vaccines that use mRNA will alter my DNA or genetic makeup.	COVID-19 vaccines will not alter your DNA.
COVID-19 vaccines cause infertility or problems during pregnancy.	There is no evidence that COVID-19 vaccines cause infertility or increase the risk of miscarriage.
COVID-19 vaccines will implant tracking microchips in people.	Vaccine injections do not contain tracking microchips.
COVID-19 vaccines will be mandatory for every Ohioan.	Ohio will not make COVID-19 vaccination mandatory.
You can get COVID-19 from COVID-19 vaccines.	COVID-19 vaccines will not give you COVID-19.
If I have recovered from COVID-19, I don't need to get the COVID-19 vaccine.	People who have recovered from COVID-19 are advised to get vaccinated.
COVID-19 isn't very serious, so I don't need to get the vaccine.	The severity of COVID-19 symptoms varies widely, and getting vaccinated can help protect you from serious illness from COVID-19.
You will get a positive COVID-19 viral test if you receive the COVID-19 vaccine.	COVID-19 vaccines will not cause you to test positive on COVID-19 viral tests.
Other vaccines, like the flu shot, will prevent COVID-19.	Only vaccines designed specifically to prevent COVID-19 will protect you from COVID-19.
Vaccines cause autism.	Vaccines do not cause autism.

Get Your COVID Vaccination Today

Protect yourself, your family and others. If you have not been vaccinated against COVID-19 or if you need a booster shot, please contact DDC Clinic at **440-632-1668** to make your appointment today. COVID vaccinations are provided at no cost to you.

Our New Website is Here

*We're pleased to announce that our brand new website is live! We invite you to visit **ddcclinic.org** and take a look:*

Our new user-friendly website is bigger and better, and features loads of information.

- Learn about our work – from patient care to research to education
- Read personal stories about our patient families
- Get to know our team of doctors
- Discover DDC Clinic's history
- See the latest DDC Clinic news and events
- Sign up for our newsletter

Our new site is mobile-friendly, so it displays and works properly on your desktop, phone or other mobile device.

Visit our new website, and learn more about us at ddcclinic.org.

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

Our friends in these Amish and Mennonite communities are holding benefit auctions to support our mission to help special needs children. Please mark your calendars and join us for food, fellowship and fun.

SHILOH
Community Auction
Saturday, July 9
Blooming Grove Auction Barn
1091 Free Rd., Shiloh, OH

SPARTANSBURG
Community Auction
Friday, August 5
215 Jefferson St.,
Spartansburg, PA

GEAUGA
Community Auction
Friday, August 26
John (Buster) Miller Farm
17719 Newcomb Rd.,
Middlefield, OH

Get details at **ddcclinic.org** or call **440-632-1668**.