DDC clinic
Center for Special Needs Children

Fall 2022
Chronicle

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

Finding Answers – Matthew's Story

This story is told by Anna, Matthew's mother.

We have a family of seven healthy children. Our sixth child, Matthew, was seven years old when I decided he needed to have his eyes checked. I had noticed for a while that his eyes looked very tired at times, and he also seemed to be looking too hard while reading a book. I made an appointment for him with an eye doctor in town. During that visit, we discovered something that we had not known before; Matthew could hardly see anything with his right eye. We also found out that he had a dislocated lens in that eye.

That doctor sent us to an eye specialist who told us that the lens in Matthew's other eye was a bit dislocated, too. We were told that he needed to see a pediatric ophthalmologist as he was a growing child.

The pediatric ophthalmologist examined Matthew, but couldn't give us any answers. He sent us to a surgeon at University Hospitals who told us he'd have to remove the lens in Matthew's right eye, and he would have to wear a contact lens afterwards. Because of our son's young age, the surgeon didn't want to put in a new lens at that time.

Before the surgeon would do the operation and put Matthew under anesthesia, he needed to know if something else was going on. He wanted Matthew to have genetic testing to see if he had Marfan syndrome or homocystinuria, as a dislocated lens can happen with these conditions.

Dr. Cruz, Matthew and Dr. Wang

The surgeon wanted us to see a University Hospitals genetic doctor, but my mom suggested we go to DDC Clinic. We took Matthew to see Dr. Wang and Dr. Cruz. Blood work came back normal for the two tests that were ordered, but the doctors weren't satisfied. They felt that our son's condition was not normal; something was causing it.

Dr. Wang and Dr. Cruz did research and custom made a genetic test for our son. The test showed that Matthew had a genetic mutation in the *ADAMTSL4* gene which

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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

Thank You for the First 20 Years!

Let them give thanks to the Lord for his unfailing love and his wonderful deeds for mankind, for he satisfies the thirsty and fills the hungry with good things. — *Psalm 107:8-9*

Here at DDC Clinic we certainly have much to be thankful for! We recently had our 22nd annual benefit auction here in the Geauga County community, and the incredible generosity we again witnessed was very humbling and fills us with gratitude. Just a few weeks earlier, the Spartansburg, PA community hosted their annual auction with huge success, and in July, the Mennonite Community in Shiloh, Ohio held their annual auction, proceeds from which they've generously shared with DDC Clinic for the last 15 years. Thank you to all for your unselfish and hard work.

We've also been blessed with very generous funding from both the local Amish community and the non-Amish communities surrounding us. We are most grateful to all our supporters for trusting and helping DDC's mission. With such generosity, we want to give back to you with services that will benefit this and future generations. I hope we can continue to put the needs of the community first, to never forget why we are here, and to focus on serving others, over personal gain.

It's now just over 20 years since Dr. Wang was hired and started treating patients. Thank you, Dr. Wang, for dedicating your life's work to our special needs children. We are very grateful that you have spent the last 20 years serving and working in our community. Lord willing, we hope to have you here for the next 20 years and longer.

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

Eli Miller

Eli Miller, Executive Director
DDC Clinic

Scrapbooks Surprise Dr. Wang

We wanted to do something special to commemorate our clinic's 20th anniversary. As a surprise for Dr. Wang, we secretly asked patient families and friends of DDC Clinic to contribute to a scrapbook we were creating. The response was overwhelming! We put their letters, cards and other keepsakes into two beautiful scrapbooks which we presented to Dr. Wang.

He was genuinely moved by everyone's kindness, and he wanted to reach out and express his gratitude to everyone who is a part of DDC Clinic. Here's what he had to say.

Dear Families and Friends,

Thank you for the scrapbooks!

What a surprise! I am overwhelmed by the kind words, compliments, and thoughts you put together and the time you spent in preparing the books. I thoroughly enjoyed each piece that you sent – the cards, pictures, handprints, paper cut-outs, drawings, Bible verses, poems, stories, and jokes … You have truly touched my heart and made me feel once again that the work at DDC Clinic is valuable.

It has been such a humbling experience to serve each child and family for the past 20 years. When a parent brings a child to DDC Clinic – from Middlefield, from another state, or another country – they put their trust in us. Thank you for the privilege you give to us.

I wanted to particularly thank our clinic's founders for their vision, our board for their leadership, the community for their trust, the countless volunteers and supporters for their contributions, and ultimately, the talented staff for their hard work and dedication in fulfilling the clinic's mission. I am indebted to all of you!

I am ending this thank you note on a personal level with a reminder from one of the parents in the scrapbook, *"Just think, you are not here by chance but by God's choosing. … You lack nothing that His grace can't give you. He has allowed you to be here at this time in history to fulfill His special purpose for this generation."*

I am in awe of this opportunity and will certainly try my best.

Humbly yours,
Heng Wang, M.D., Ph.D.

Benefit Auctions a Huge Success

Each year, our friends in Amish and Mennonite communities hold benefit auctions in support of our work with special children. The dollars raised from these auctions are crucial, enabling us to provide life-changing diagnoses and treatments to children affected by rare genetic disorders.

This year's auctions were our most successful ever. We thank our friends in Shiloh, Spartansburg and Middlefield for hosting these benefit auctions and for their generous donations of goods, time and labor. We're truly humbled by the continued support of so many.

Shiloh Community Auction

We're grateful to the auction organizers and supporters who attended the July 9th Shiloh Community Auction. This yearly auction benefits both DDC Clinic and the Clinic for Special Children in Strasburg, Pennsylvania. Proceeds from this auction have grown every year since its start in 2007. Many thanks to our friends in Shiloh.

Spartansburg Community Auction

We also thank our friends in Crawford County, Pennsylvania for raising funds for our clinic at the 11th annual Spartansburg Community Auction on August 5th. This out-of-state auction is held every year to help support our mission as a number of families from the Spartansburg area travel to DDC Clinic for their children's care. Among the many items donated to the auction were handmade quilts, furnishings and farm-fresh local produce.

Geauga Benefit Auction

Thanks to the amazing generosity of more than 2,000 attendees and the auction committee's hard work, the 22nd Annual Geauga Benefit Auction held on August 26th was a record-breaking success. Auction items included a flagstone patio with fire ring, custom kitchen cabinets, timber frame pavilion, custom two-car garage and handcrafted quilts.

Understanding Genetics

GENETICS and EYE DISEASES

Your eyes are as unique as you are. Your eye color, whether it's blue, brown or any other color, is influenced by your genetics. The genes that are passed on to you from your parents not only determine the color of your eyes, but they can also affect the health of your eyes.

For the past 30 years, our knowledge and understanding of eye diseases have increased substantially, specifically for those diseases influenced by genetics. While the genes and genetic variations that determine the color of our eyes do not cause eye disease, other genetic variations do.

In the Plain community, these are some of the conditions which we have seen which can cause eye diseases.

- **Jalili syndrome** is a change in the gene called *CNNM4* that causes progressive vision loss (cone-rod dystrophy) and teeth abnormalities (amelogenesis imperfect).
- **Ectopia lentis** is a disorder in the lens of the eye (dislocated lens) caused by a change in the *ADAMTSL4* gene.
- **Retinitis pigmentosa** is a disorder caused by a change in the gene called *SCAPER* that leads to vision loss and intellectual disability.
- We found changes in two genes, namely *TYR* and *OCA2,* related to a condition called **ocular albinism** in members of our community. This disease is characterized by severely reduced pigmentation of the iris, which causes very light-colored eyes and significant problems with vision.
- **Myopia (nearsightedness) and deafness** can be caused by a change in the *SLITRK6* gene.
- Other genetic conditions that contribute to significant vision problems include **Cohen syndrome, GM3 synthase deficiency** and **Leigh syndrome.**

About a year ago, genetic testing performed by our onsite molecular diagnostics laboratory revealed that one of our young Amish patients had ectopia lentis caused by a mutation in the *ADAMTSL4* gene. *(See the story on page 1.)* It was the first case we had ever seen. Ectopia lentis is an autosomal recessive condition that equally affects males and females. "Recessive" means that two copies of the mutated gene (one from each parent) must be present to cause the disorder.

In the case of our patient, the boy's parents passed the gene mutation on to him. In a family where the parents don't have the disease, but both are carriers (each parent having one copy of the mutated gene), there's a 25% chance that their children will inherit the condition.

The *ADAMTSL4* mutation in the Amish is very old and pre-dates the Amish coming to America – and it is the same exact disease seen in Northern Europe and Scandinavia. So this disease is not an "Amish disease," but rather a European disease.

People with this disease can have very dramatically different problems. Some will have only mild symptoms requiring glasses. Others will have one or both of their lenses "pop out of place" and need surgery to replace them, and a few will have abnormally shaped pupils at birth.

Since diagnosing our young patient with ectopia lentis, we've found others in our community affected by the same condition. We can now test for it through our Genetic Awareness Panel (GAP) enabling us to detect the condition.

Having the right diagnosis can prevent unnecessary and costly additional testing and imaging. More importantly, the right diagnosis enables a child's eye doctors to take their young patients to surgery with confidence and helps them to monitor for long-term complications from an early age.

CLOSING THE GAP

Helping Families Affected by Pediatric Cardiomyopathy

At DDC Clinic, we're committed to furthering personalized medicine and creating greater awareness of genetic diseases. Our efforts help to "close the gap" by building bridges between the Plain community and the healthcare system.

One of the ways we've helped to close the gap is through our affordable Genetic Awareness Panel (or GAP) which can simultaneously test for more than 160 rare genetic conditions found in the Amish population.

We've also worked to narrow the gap through collaborations with major medical centers, researchers and pharmaceutical companies, such as Tenaya Therapeutics.

DDC Clinic was the first institution to be chosen to participate in Tenaya's five-year global multi-center natural history study designed to help better understand the impact of *MYBPC3* genetic mutations on heart disease, including hypertrophic cardiomyopathy, in children.

Our clinic was a logical choice for this cardiomyopathy study. For more than 20 years, we've provided patient care and support to Amish families from Ohio and surrounding states whose children have been affected by *MYBPC3* gene-related severe infantile cardiomyopathy.

Our ongoing participation in this important study has enabled us to learn more about cardiomyopathy, one of the most common causes of sudden death in adults and children.

"We're always searching for answers and sharing the knowledge we gain with our patient families. One of the ways we do this is through family gatherings, such as the one we held this past summer for families touched by cardiomyopathy," says Dr. Wang, DDC Clinic Medical Director.

Cardiomyopathy Family Gathering

On July 23 of this year, we had the pleasure of hosting the Hypertrophic Cardiomyopathy Family Gathering at DDC Clinic. Amish families who lost children due to *MYBPC3* gene-related cardiomyopathy came for this day-long educational program which provided families with an opportunity to share, learn and support one another in a comfortable, familiar setting.

Families learned about the genetics of cardiomyopathy and its diagnosis, treatment options and gene therapy. They also heard about our clinic's participation in Tenaya's cardiomyopathy natural history study and future clinical trials. In addition, families were offered free targeted *MYBPC3* carrier testing performed by our onsite molecular diagnostics laboratory.

The day's agenda featured discussions, Q&A sessions and presentations by DDC Clinic doctors and staff, including Eli Miller, Executive Director; Dr. Heng Wang, Medical Director; and Dr. Vince Cruz, Geneticist and Internist.

Guest speakers included Dr. Kenneth Zahka, Pediatric Cardiologist at Cleveland Clinic and Dr. Nathaniel Robbins, Pediatric Cardiologist at University Hospitals as well as representatives from Tenaya Therapeutics including Dr. Laura Robertson, Executive Medical Director and Wendy Borsari, Patient Advocacy.

"By working with patient families through gatherings such as this, performing targeted genetic carrier testing and increasing awareness of risk for genetic diseases," says Dr. Wang, "we're moving one step closer to narrowing the gap between the Plain community and the medical community."

Has your family lost children or had a child with a heart transplant due to *MYBPC3* gene-related cardiomyopathy?

DDC Clinic has created a new Cardiomyopathy Newsletter for families affected by this disorder. To be added to the mailing list, please contact us at 440-632-1668.

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affects the part of the eye that holds the lens in place. The two doctors had never seen a case like this before, but they had learned there were rare cases in Germany, Norway and Sweden. They got information about how the condition was treated there.

The DDC Clinic doctors told us both parents have to be carriers in order for the child to have the condition. They also said that one in four of our children could have it. We had our six other children tested, and we learned that none of them had Matthew's condition, but they were all carriers. The test results and other information were sent back to the eye surgeon, and we were finally able to move forward.

Matthew had the surgery to remove the lens in his right eye. He had a two-month recovery, and then he got a contact lens. He also wore a patch over his left eye (his good eye) to help develop better vision in his right eye. His left eye continues to be watched by doctors, as that lens could dislocate more. For now, Matthew has excellent vision in his left eye, and we hope it stays that way.

DDC Clinic helped our family to find answers. We highly recommend Dr. Wang and Dr. Cruz to anyone who needs genetic testing. We wouldn't know what we now know if it weren't for them.

Patchwork Benefit Raises $110,000

Thanks to our generous supporters, this year's Patchwork Benefit was a major success, raising $110,000 to support DDC Clinic's mission.

"This was our most successful Patchwork Benefit ever," says Patti Gallagher, the event's organizer and DDC Clinic's Operations Director. "We're so grateful to our supporters who make it possible for us to make a real difference in the lives of special children."

This year's event was a special celebration commemorating DDC Clinic's 20-year journey of faith, hope and caring. More than 200 guests enjoyed hors d'oeuvres, drinks, dinner, auctions and the ever-popular Dessert Dash.

The highlight of the evening was a presentation by current and former board members, JoAnn Brace, Gordon Safran and Tom Stone. Each took part in recounting DDC Clinic's remarkable 20-year history, including our clinic's humble beginnings, the hiring of Dr. Wang, our significant achievements over the years and our path to becoming the world-class medical facility we are today.

This year marks the first time that our Patchwork Benefit was held since 2019, as the event was canceled in 2020 and 2021 due to COVID. This year, our loyal Patchwork supporters returned in full force, eager to do their part to help our mission.

"It's wonderful to know that so many people support DDC Clinic and that they continue to put their steadfast belief, faith and trust in the work we do," says Patti. "We look forward to seeing everyone again at next year's Patchwork Benefit on June 17, 2023."

Welcome, Bea

Meet our new staff nurse, Bea Torres-Fults, who joined DDC Clinic in May of this year. Bea came to us from the Neonatal ICU at MetroHealth Medical Center, where she worked with newborn infants and developed her love of caring for the pediatric population.

At DDC Clinic, Bea cares for patients from infancy to adulthood. Bea is very excited to share her knowledge and passion for nursing with our patient families, and she enjoys helping those in need.

Reflections on 2021

When we reflect upon another year, we're always filled with gratitude for the support of our many friends whose generosity has helped us to continue our most privileged work.

As we prepared this 2021 financial report, we were reminded of everyone who helped spread the word about our services, participated in our benefit auctions, collaborated on special projects or helped to support our mission in a myriad of other ways.

2021 was a year of achievements and challenges for our clinic. We were very pleased to welcome Eli Miller as our new Executive Director. Eli, who is Amish and a lifelong resident of Geauga County, brought a much-needed perspective to our leadership team and helped to further deepen our ties to the Plain community.

We also welcomed Dr. Vince Cruz as our clinic's second physician. With Dr. Cruz onboard, we expanded our scope of services to include continuity of care, enabling our patients to receive highly personalized care as they mature into adolescence and adulthood.

Despite challenges related to the COVID pandemic, we saw 534 patient families in 2021 and administered 1,134 childhood vaccinations. Our Patient Assistance Program helped families in need by distributing 1,045 cases of formula, 450 boxes of biliary supplies, 76 cases of feeding bags and 120 feeding buttons.

COVID also presented us with financial challenges. Due to the pandemic, we canceled our Patchwork Benefit, a major fundraiser, for the second year in a row. In its place, we hosted a special thank you event for a small group of donors and supporters who stood by us throughout that unprecedented and difficult time.

On another front, we saw important developments related to our research endeavors, specifically our multi-year partnership with Regeneron Genetics Center. Working together to study undiagnosed rare conditions in the Amish, we identified unknown gene mutations and uncovered the underlying causes of rare diseases affecting members of our community.

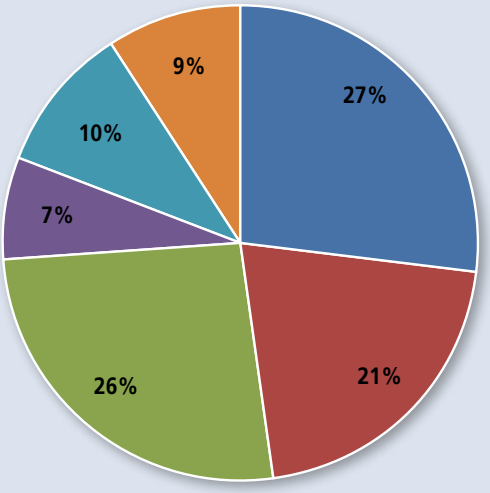
We also continued our decade-long study of TMCO1 Defect Syndrome, and shared our research findings in a scientific article published in *Frontiers in Genetics.*

Our continued progress in patient care, research and education in 2021 was only made possible by the support of our many friends. We couldn't have done it alone. Working together, we were able to provide hope to parents and life-changing care to their special needs children.

Thank you for believing in our mission. We're truly blessed to have so many good friends.

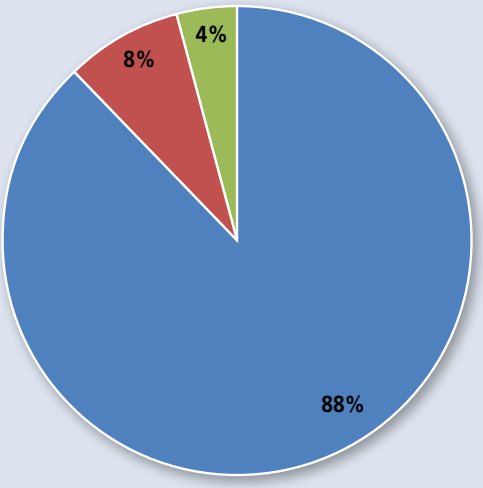
2021 Financial Report

Total Revenue $1,996,239



Contributions	$536,647
Fundraisers	$410,856
Clinical Services	$515,814
Research Contracts	$145,801
Miscellaneous Income	$204,142
Individual Giving	$182,979

Total Expenses $1,467,768



Program Services	$1,289,523
Fundraisers	$116,706
Management & General	$61,539

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

Celebrating 20 Years of Faith, Hope and Caring

We've been making a difference in the lives of special needs children for 20 years. Since our clinic opened its doors, we've helped more than 1,600 patient families.

But we couldn't have done it alone. It's people like you who have made our work possible. When you support our clinic, you help children affected by rare genetic disorders get the life-changing and life-saving medical care they need.

Will you consider making a tax-deductible donation to help a child live a better, healthier life?

To make a gift, use the donation envelope inside, visit **ddcclinic.org** or call **440-632-1668**. We thank you for your support.