DDC clinic
Center for Special Needs Children

Fall 2019
Chronicle

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

Amish Genetic Disease Panel Update

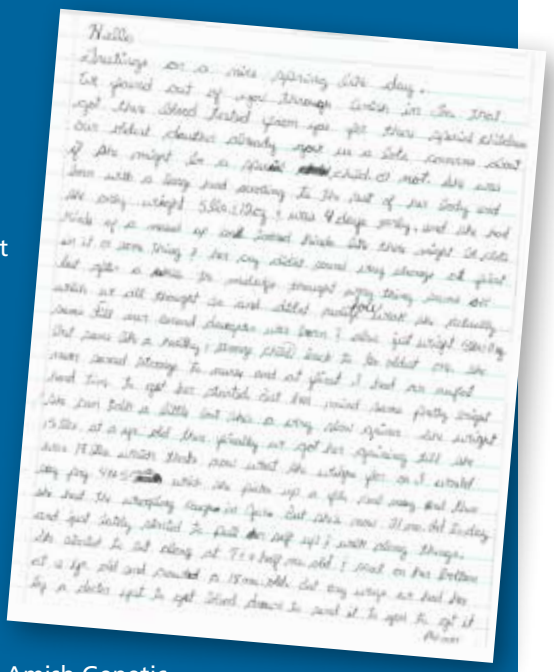
Last summer we announced the development of a groundbreaking diagnostic tool that simultaneously tests for over 120 rare conditions.

Through this panel, we have been able to provide answers to many families affected from undiagnosed genetic conditions. The panel has reached families throughout the Midwest, and we have been able to diagnose patients affected by a variety of conditions including GM3 Synthase Deficiency, Byler Disease, and Troyer Syndrome, allowing them to seek specific treatment. Since we have launched the Amish Genetic Disease Panel we have provided testing to nearly 150 families.

DDC Clinic Molecular Diagnostics Laboratory has been able to diagnose children who were experiencing significant medical problems in Ohio, Kentucky, and Tennessee. These diagnoses provide answers and allow the patients to receive specialized care.

Additionally, the Amish Genetic Disease Panel has identified risk factors for nearly half of individuals who have been tested. Most commonly, we have been able to identify individuals who are at increased risk of developing a blood clot or a heart condition as an adult. This information is important for patients, providing awareness of health risks they may experience in the future.

Troyer Syndrome Story



Several months ago, we received a phone call from a concerned Nurse Practitioner in Tennessee. The Nurse Practitioner had visited a family in rural Tennessee who had just had a baby. When the Nurse Practitioner met with the family, she noticed that several of the older children seemed to have developmental delays and looked different from their siblings. The family said that they had met with many doctors to try to figure out what was going on with their children, but no one had been able to provide them with an answer. The Nurse Practitioner called our lab for advice.

Our genetic counselor suggested that the Nurse Practitioner offer the Amish Genetic Disease Panel to the family and test one of the affected children to see if we could find a diagnosis.

Fortunately, our panel was able to provide an answer for this family. The child was diagnosed with Troyer Syndrome, a genetic condition in which children have trouble walking, difficulty speaking, weakness, trouble dealing with their emotions, and intellectual disability. Because DDC Clinic was able to provide the family with a diagnosis, they could now receive specific services that could help their children lead a happier and healthier life.

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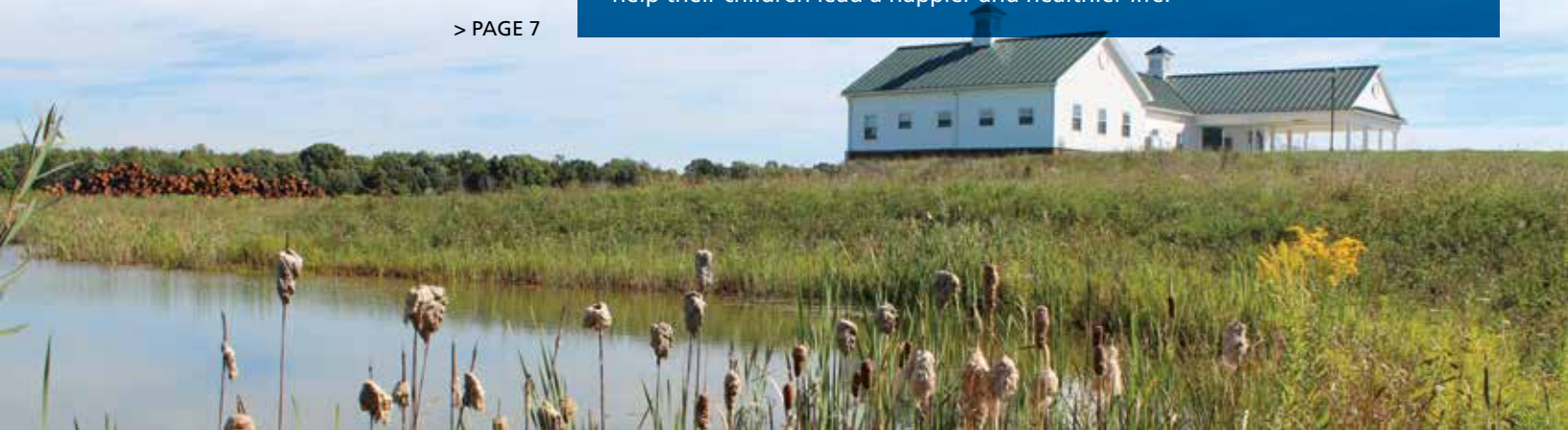

About DDC Clinic

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 1,000 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 70 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.

DNA-Themed Quilt Created for DDC Clinic Lab

The ladies of DDC Clinic's Quilting and Craft Club created a quilt, featuring DDC Clinic's signature blue and brown colors. The bold pattern represents DNA strands, the genetics-themed quilt was designed for use as a backdrop for our booth displays at meetings attended by our lab staff.

The Quilting and Craft Club works on a variety of projects, focusing mostly on creating hand-stitched quilts. The group, who donate quilts every year to benefit auctions, is currently working on a quilt that they plan to donate to our Geauga Benefit Auction in October.

Now in its eighth year, the Quilting and Craft Club meets the first Wednesday of each month from 9 a.m. to 1 p.m. at DDC Clinic. The group always looks forward to welcoming new faces, and they invite any interested ladies from the community to bring a lunch and join them for some quilting and visiting.

Ladies' Lunch and Get-Together

More than 80 ladies from the Geauga and Holmes County settlements gathered in the community room at DDC Clinic on Wednesday, April 24 for lunch and socializing. The Mom's Support Group from the Geauga settlement provided a Haystack lunch, and the ladies from Holmes County provided assorted desserts. The Holmes County group also spent part of the day touring our local special needs schools and the new Sunshine Training Center adjacent to our clinic.

Looking to Volunteer?

You can make a difference. Do you know anyone that could volunteer to assist with a few "chores"? We could use some help with power washing our building, weeding and trimming all of our garden areas every other week, planting a few perennial flowers and refinishing our front doors. Contact Patti at 440-632-1668 for more information.

THANKS TO OUR *Patchwork* SUPPORTERS

We offer our warmest gratitude to those who joined us at our annual Patchwork Benefit on Saturday, April 13. Thanks to the generosity of our supporters, we raised $90,000 in support of our mission.

This year's event was our 19th annual Patchwork Benefit, and the 20th anniversary of the founding of DDC Clinic. Three hundred guests gathered to celebrate 20 years of our clinic's achievements, ranging from life-changing patient care to pioneering genomics research to the sharing of groundbreaking discoveries.

The evening's festivities began with hors d'oeuvres and live piano music. A selection of Amish hymns preceded dinner, a delicious menu featuring grilled sirloin, shrimp, scallops, penne pasta, mini meatballs, tacos and more.

After dinner, Dr. Heng Wang, our clinic's medical director, presented a program about our work with Cohen Syndrome patients and spoke about one family's personal story.

Guests enjoyed the excitement of a live auction which included items such as an Amish quilt, Playhouse Square tickets, a Big Green Egg barbecue cooker and an Amish wedding dinner for 10.

Our popular Dessert Dash, basket raffle and bake sale rounded out the memorable evening.

Proceeds from this year's Patchwork Benefit will help further our clinic's mission, improve the quality of life for children with special needs and provide support services to families.

Save the Date

We hope to see everyone again at our next Patchwork Benefit on April 18, 2020.

THANK YOU TO OUR PATCHWORK SPONSORS

We're grateful to the businesses and individuals who supported this year's Patchwork Benefit.

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Annie Xin

On July 20, 2019 over 180 family members visited DDC Clinic for a gathering of patients with Byler Disease.

Speakers that joined us for the day included DDC Clinic staff, Dr. Heng Wang, Sarah Ossler, genetic counselor and Valerie Sency, registered nurse. Additional speakers included Dr. Vera Hupertz, Pediatric Medical Director of Hepatology and Liver Transplantation Cleveland Clinic; Dr. Kyle Soltys, Associate Professor of Surgery, Children's Hospital of Pittsburgh; and Asta Slechticovas from the Ronald McDonald Care Mobile, University Hospitals Cleveland.

A chicken barbecue was served for lunch and everyone enjoyed visiting with one another the rest of the afternoon. Many were overheard saying that the time went so fast!

We would like to acknowledge how grateful we are to Albireo for the sponsorship of this gathering.

October 12, 2019 is the Geauga Benefit Auction!

Enjoy good-natured bidding, delicious food and warm fellowship. The auction held on an Amish farm (17719 Newcomb Road, Middlefield, Ohio) begins with breakfast served at 5:30am until the auction starts at 9:00am. If you don't make it that early, not to worry! Plenty of food is available throughout the day.

A basket raffle might pique your interest, or perhaps some goodies at the Amish bake sale. Enjoy lunch and take home a homemade pretzel!

Bid on a variety of auction items including quilts, furniture, tools, horses, buggies, firewood and much more. You will never know what interesting items you might find.

Again this year, we will have a 24' x 32' garage to be built on your prepared site. Another popular item is a 16' x 20' timber frame pavilion.

Raffles this year give you a choice of a complete bedroom set in your choice of style and finish from Cherry Valley Furniture (value up to $4,500) and also a Hunting Blind from Buckeye Structures (value up to $2,000). Tickets will be sold for $10.00 each or 6 for $50.00 and are available at DDC Clinic. New items are being added all the time, so stay tuned.

The **13th Annual Shiloh Mennonite Community Benefit Auction** was held on Saturday, July 13 in Shiloh, OH and the **7th Annual Spartansburg Benefit Auction** was held on Friday, August 9 in Spartansburg, PA. Both events were blessed with beautiful weather and had large crowds. The Shiloh auction raised $37,000 and the Spartansburg auction raised $70,000. We are grateful to the communities who hold these annual benefit auctions and the many community members and volunteers who lend a hand to make them possible.

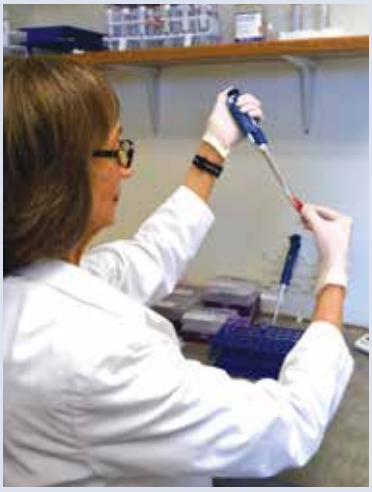
The money raised from our community benefit auctions allows us to provide life-changing diagnoses and treatments for special needs children and their families.

Will you help us help a special child?

DDC Clinic provides highly personalized and comprehensive care in a medical home dedicated to patient-centered research. Our commitment is to find answers for families and support them with the best of care for their children - regardless of ability to pay. We are a place of faith, family, hope, and compassion.

As a community-supported clinic, we would not be here without your blessings, trust, and support. The majority of DDC Clinic's budget comes through donations, and your generosity helps us save lives, prevent disabilities, and decrease children's needless suffering.

- **Life changing services are provided to families for significantly less than our cost. A one-hour office visit is just $55, and many of our genetic tests cost self-pay families just $75.**
- **Our clinic's Patient Assistance Program provides low and no cost medicines, specialized formulas and medical supplies, saving the local community over two million dollars last year alone.**
- **Rapid diagnoses and proactive care eliminate "diagnostic odysseys" for families and help prevent needless and costly hospitalizations.**
- **We conservatively estimate that every donated dollar leverages eight to ten in saved healthcare expenses in return – an estimated 11-14 million dollars in medical savings annually.**



Individually, the disorders we treat are indeed rare, but diagnoses and care of special children should not be. Thank you for helping us make a difference!

Understanding Genetics

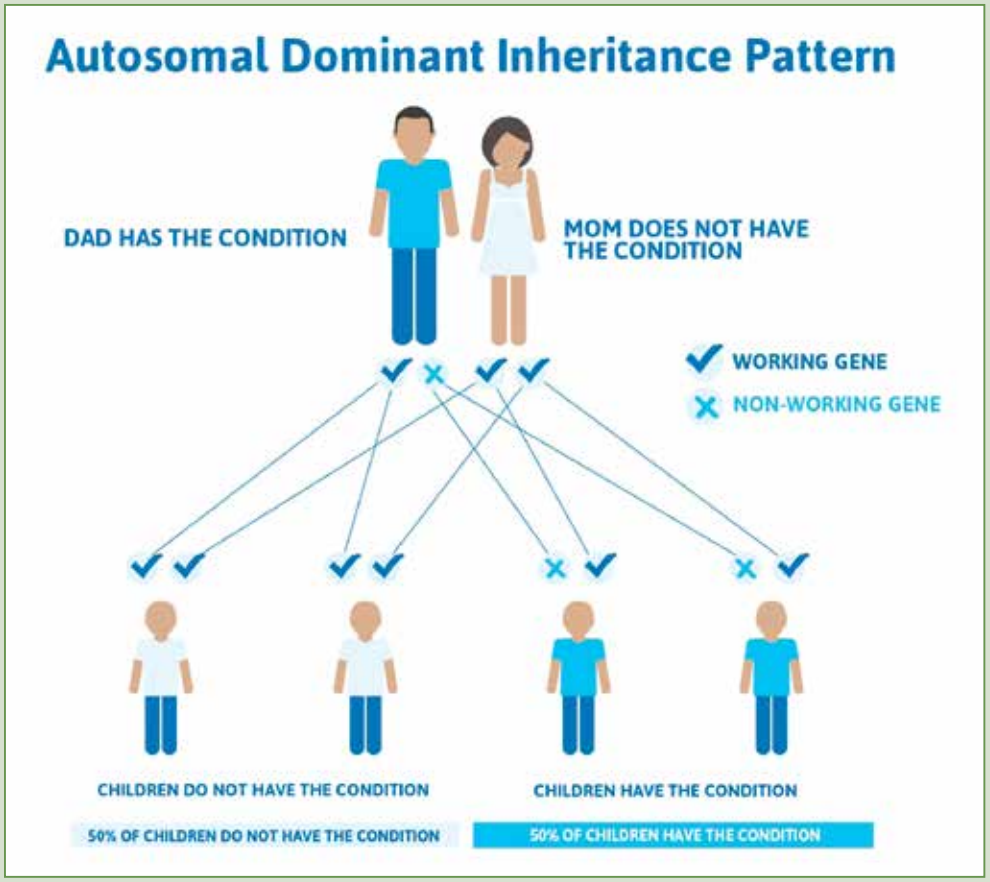
Inheritance Patterns

Genetic conditions run in families in different patterns. Many, but not all of the disorders DDC Clinic diagnose are recessive. This means that for an individual to have the disorder, they must inherit two copies of the genetic change, one from each parent. With each birth, parents who carry the same genetic change have a 25% chance of having a child with the disorder. We reviewed recessive inheritance in our Fall/Winter 2018 newsletter.

This article highlights a different type of inheritance: the dominant inheritance pattern (sometimes called autosomal dominant). The article also explains penetrance; a concept that individuals who inherit a disorder can sometimes show minor or no symptoms at all, while other individuals who inherit the same disorder may be severely affected.

Dominant Inheritance

Dominant inheritance is when a person only needs to have one genetic change to be affected by the disorder. Affected individuals have a 50% chance of passing their genetic change to their children. The chance that a child will not inherit the genetic change is also 50%. This does not mean that if you have a dominant genetic change, that half of your children will have the disease. It is possible that all of your children could inherit the disease, or none of them. Each child has his or her own 50% chance of inheriting the genetic change, regardless of whether its siblings have or have not inherited the genetic change. Sometimes, many individuals in a family with a dominant condition may inherit a disease, and other times, only a few will inherit the disease.



Penetrance

Penetrance refers to the chance that an individual with an inherited condition will show symptoms of the disorder. In some conditions, inheriting a genetic change does not necessarily mean you will have symptoms associated with the disease. Some genetic changes may run in a family, but not every family member will show symptoms; this is reduced penetrance. One family member who has a genetic change may have many health problems, while another family member with the same genetic change may have few health problems.

Doctors and researchers do not yet fully understand how this happens, but suspect the interaction of genes, environment, and lifestyle may determine how many symptoms a person may have. This can be challenging for doctors and frustrating for patients and families, as it is difficult to predict if an individual will show symptoms of the disease based solely on their genetic status.

Hypertrophic Cardiomyopathy: A Dominant Condition with Reduced Penetrance

It is possible for a genetic disorder to be dominant and show reduced penetrance. An example is hypertrophic cardiomyopathy, a condition when the muscles of the heart thicken, making it harder to pump blood through the body. Hypertrophic cardiomyopathy can cause shortness of breath, chest pain, or problems in the heart's electrical system, resulting in life-threatening abnormal heart rhythms (arrhythmias). Typically, symptoms begin in adulthood.

In the Amish population, genetic changes in the gene *MYBPC3* are the most common cause of hypertrophic cardiomyopathy. Hypertrophic

cardiomyopathy is an autosomal dominant condition. This means that if you have hypertrophic cardiomyopathy, there is a 50% chance that you will pass the condition on to each of your children.

Hypertrophic cardiomyopathy also has reduced penetrance. This means that individuals who inherit an *MYBPC3* genetic change may or may not have symptoms. Doctors are unable to predict if a person will have symptoms based on their genetic status alone. Because of this, doctors monitor patients who have an *MYBPC3* genetic change to see if they are developing symptoms before they become life threatening.

If you or a family member has hypertrophic cardiomyopathy, it is helpful for other family members to get genetic testing to see if they also carry an *MYBPC3* genetic change that would put them at risk of developing hypertrophic cardiomyopathy. If you or a family member already know that you carry a genetic change in the *MYBPC3* gene that causes hypertrophic cardiomyopathy, you should consult with your physician and follow their recommendations.

In the Amish community, sometimes a child inherits two genetic changes that cause hypertrophic cardiomyopathy, one from each parent. This causes a very severe form of cardiomyopathy. This article is NOT referring to the severe type of hypertrophic cardiomyopathy seen in children.

DDC Clinic Hosts Genetic Counseling Students

DDC Clinic has recently entered a partnership with the Genetic Counseling Master's Degree Program at Case Western Reserve University. Our laboratory has agreed to host two genetic counseling students each spring semester as part of their training. The rotation is to teach students about genetic counseling in the laboratory setting, as well as expose them to disorders specific to the Amish community. This spring two students, Molly Ford and Emily Creque, each spent four weeks in the laboratory with our genetic counselor, Sarah Ossler.

Emily Creque

Molly Ford

Hemophilia Education for the Plain Community

DDC Clinic teamed up with the Northern Ohio Hemophilia Foundation and University Hospitals to present an educational program on hemophilia, a rare disorder that affects the body's ability to make blood clots. Eighty people attended a dinner and program held at our clinic on Monday, April 15. Among the presenters was Sarah Ossler, DDC Clinic genetic counselor, who gave a talk, entitled *"The Genetics of Hemophilia."*

On Wednesday, July 31, Dr. Lalitha Nayak along with Susan Hunter and Jan Martin from the Hemophilia Treatment Center, University Hospitals Cleveland Medical Center had 20 women from the local community come for dinner and information on Hemophilia and what effect it has on your life. A discussion directed by the women in attendance followed the presentation.

Amish Genetic Disease Panel Update

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DDC Clinic was also able to present preliminary findings from the Amish Genetic Disease Panel at the national American College of Medical Genetics and Genomics (ACMG) meeting in Seattle, Washington this April. Many providers were excited about the results of our testing, and showed interest in sharing the testing options with their patients. The Amish Genetic Disease Panel has proven to be an important tool and is a great example of personalized medicine in action.

Finding Answers for Families using the Amish Genetic Disease Panel

All are welcome!

October 25th, 2019
4:00 pm-5:00 pm

Holmes County
Training Center
8001 TR 574
Holmesville, OH 44633

Call Sarah at 440-632-5532 for more information.

DDC clinic
Center for Special Needs Children

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