

President's Letter

Dear Friends,

As you settle down with this "A Year in Review" issue of the DDC Clinic Chronicle and perhaps a cup of tea, relaxing from the hustle and bustle of the holiday season, you will find much has changed at the clinic and yet, much remains the same.

First and foremost, the need – our reason for being – is still very real. The clinic remains committed to its mission of enhancing the quality of life for special needs children with metabolic and inherited disorders through early diagnosis and treatment, research, and education. We continue to strive toward our vision of providing the highest quality care and effective research for children with complex medical needs, fully supported by our local community and benefiting from an ever-widening network of support.

This year our ever-widening network of support included the Johnson & Johnson Community Health Care Crystal Award (the award and grant money are explained on page 1). It was an honor and privilege to attend the awards ceremony in Washington, DC with Dr. Wang and several other board members. Getting to know the other recipients of the award was awe-inspiring; these remarkable people and their organizations are truly making our world a better place. In addition, the workshops that were part of the weekend provided excellent ideas that will help us further improve our "medical home" for special needs children.

Dr. Wang has worked diligently at increasing our network of support (see the recent publications and presentations listed on page 2) and for all his work was recognized with the Special Achievement Award from the American Academy of Pediatrics. The fully functional research laboratory at DDC Clinic is increasing our support with significant discoveries; Dr. Xin has established DNA tests for 20 genetic diseases in this area such as Troyer's syndrome and Amish brittle hair syndrome (see the article on page 2 to learn more about this disease).

In a year with many changes, much stays the same...including our need for your support. This year we've rejoiced in your continued support, your trust and belief in the DDC Clinic. On behalf of the Board of Directors and the clinic staff, thank you.

Darla C. Klein
President, Board of Directors



DDC Clinic

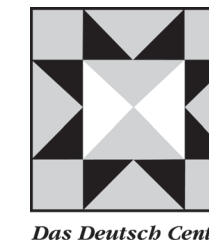
for Special Needs Children

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Enhancing the quality of life for special needs children with metabolic and inherited disorders through early diagnosis and treatment, research and education.

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DDC Clinic

for Special Needs Children

CHRONICLE

2006



INSIDE...

Publications and Presentations 2
Amish Brittle Hair Syndrome 2
Patchwork Benefit 3
Amish Benefit 4
President's Letter 5
Fundraising & Development 6

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Welcome

2006 brought about many changes to DDC Clinic. Two new directors have joined our Board this year. Eli Miller of Burton and Chuck Fowler of Lyndhurst are the most recent additions to our Board of Directors. We have also welcomed new staff to the clinic this past year. Patti

Gallagher of Burton is the Office Manager, Carol Troyer of West Farmington is the administrative assistant and Maria Burkhammer of Chardon is the new Registered Nurse. We thank all of you for your dedication and commitment to DDC Clinic.

Standing Left to Right: Heng Wang, MD, PhD, Baozhong Xin, PhD

Sitting Left to Right: Patti Gallagher, Carol Troyer

Not Pictured: Maria Burkhammer.



DDC Clinic for Special Needs Children Honored with Johnson & Johnson Community Health Care Crystal Award

The DDC Clinic for Special Needs Children received a Johnson & Johnson Community Health Care Crystal Award and a two-year \$150,000 grant at an awards ceremony on Monday, April 24, 2006 at which Senator Patty Murray (D-Washington) presented the 2006 Community Health Care Program Awardees. The Crystal Award recognizes organizations for their commitment to providing the medically underserved with innovative community health-based services.

Through funding from the Johnson & Johnson Community Health Care Program, the DDC Clinic will expand its current services to include an on-site rehabilitation program, offering physical, occupational, and speech therapies, and case management services. Additional support for the Amish families will include transportation to and from the clinic. This initiative will benefit Amish children who would not otherwise receive treatment for their conditions.

3rd Annual National Cohen Event

The 3rd Annual National Cohen Family Event was held this year in June at the Red Maple Inn in Burton, Ohio. What a warm and inviting setting for our informal day of questions and fellowship. Families traveled from Canada, Mississippi, Maine, Pennsylvania, Iowa as well as local families

from our community.

Friday was informational for parents with presentations from Dr. Xin, Dr. Wang and Leah Nye. Saturday was a family picnic at the Burton Historical Society and then our good-byes. A great time of information sharing and fellowship for all that attended.

Fundraising and Development

As the work of fundraising is never done, DDC staff, volunteers, and donors have zealously rallied to the cause of raising much-needed dollars to support the Clinic's patient care, research, and education programs. Together this year, we have rejoiced:

★ ...when the third year matching goal of \$114,175 was reached for the Robert Wood Johnson Foundation grant. Special thanks to our major supporters The Cleveland Foundation, The Michael Pender Fund, the Evie & Gordon Safran Philanthropic Fund, Susan and John Turben Foundation, James Schill, Johnson & Johnson, the State of Ohio Childcare Settlement Grant, Gold Key Processing Ltd., the Holmes County Association for Handicapped Citizens, and the Amish Traditions & Treasures event.

★ ...when the Annual Patchwork Benefit Auction netted over \$66,583 with special thanks to co-chairs Sue Patterson and Mary Ruth Meyer, along with their entire committee. Hurray for the ambiance, dinner, and fantastic auction items!

★ ...when Geauga County Commissioners Craig Albert, Mary Samide, and Bill Young awarded DDC Clinic a \$20,000 County Bloc Grant for DNA testing. These funds will be doubled by the matching grant.

... and we have rejoiced each and every time we received a donation, no matter the amount, from supporters who genuinely care about our children and the work we do.

We rejoice in the knowledge that our donors have been so generous, so compassionate, and so willing to invest in our future. Thank you to each and every one of you.

If you would like to rejoice with us by making a donation, or learn more about planned giving options, please call the clinic office at 440.632.1668 so we may talk to you personally.

Annual Report Mailed ~ The annual report for DDC Clinic was mailed in November to all donors.

While we strive to have the most accurate information to list our donors, we acknowledge that we are indeed human and may make errors. We humbly apologize for any omissions that may have occurred including **Lander Circle Kiwanis Club**.

Our annual report also reported 200 patient care hours provided free of charge. We were actually able to provide 2000 hours through support from our Children's Fund.

I will gladly support DDC Clinic. Enclosed is my check.

Please make the donation in Memory / Honor of:

Name _____

Occasion _____

Please acknowledge my gift to:

Name _____

Address _____

My Name _____

Address _____

City _____ State ____ Zip _____

Home Phone _____

Email _____

Make checks payable to:

DDC Clinic
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DDC Clinic for Special Needs Children is a 501 (c) (3) organization. Donations are tax deductible to the extent permitted by law.

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Research Publications and Presentations

DDC Clinic has made enormous progress in advancing the understanding of many diseases through our patient care and research. Here is a list of recent research publications and presentations by DDC Clinic and collaborators.

- Prolidase deficiency and biochemical assays used in its diagnosis. *Analytical Biochemistry* 349: 165-175; 2006.
- A nonsense mutation of PEPD in four Amish children with prolidase deficiency. *American Journal of Medical Genetics* 140A: 580-585; 2006.
- Cohen Syndrome in: GeneReviews at GeneTests: Medical Genetics Information Resource [database online]. Copyright, University of Washington, Seattle, 1997-2006. Available at <http://www.genetests.org> since August 2006.
- Etiology of vision loss in ganglioside GM3 synthase deficiency. *Ophthalmic Genetics* 27:89-91, 2006.
- GM3 synthase deficiency: A disorder of ganglioside biosynthesis presenting as an infantile epilepsy. Presented in 34th Annual Meeting of the Child Neurology Society in Los Angeles, California, September 2005. *Annals of Neurology* 58: S102, 2005.
- Community-based medical home for patients with complex genetic disorders – A model to follow? Presented in the American Society of Human Genetics 55th Annual Meeting in Salt Lake City, Utah, October 2005.
- Initiation and preliminary analysis of National Cohen Syndrome Database. Presented in the American Society of Human Genetics 55th Annual Meeting in Salt Lake City, Utah, October 2005.
- Two new metabolic disorders in Amish community. A talk presented in Cleveland Citywide Genetics Meeting at University Hospitals in January 2006.
- Etiology of vision loss in GM3 synthase deficiency. Presented in The Association for Research in Vision and Ophthalmology Meeting in Fort Lauderdale, Florida, April 2006. *Investigative Ophthalmology & Visual Science E-Abstract* 4615, 2006.
- Medical home for patients with complex genetic disorders in the Amish community - A model to follow? Presented in International Symposium on Genetics and Healthcare in Amish and Mennonite Communities in London, Ontario, April 2006.
- Study genetic disease without electricity - Genetic disorder care and research in the Amish community. Pediatrics Grand Rounds presented in Le Bonheur Children's Medical Center, Memphis, Tennessee, May 2006.
- Building a bridge between Amish culture and modern molecular medicine, Pediatric Grand Rounds presented Cleveland Clinic Children Hospital, September 2006.
- Patient-oriented research in children with complex genetic & metabolic disorders: success, challenges & opportunities for a primary care facility. Presented in Building Bridges - Care of the Medically Fragile Child National Conference in Louisville, Kentucky, September 2006.
- Building a bridge between Amish culture and Genomic Medicine, Grand Rounds presented Hamot Medical Center - Main Campus in Erie, Pennsylvania, December 2006.

Amish Brittle Hair Syndrome

In the early 1970s, a few physicians first described a brittle hair syndrome in Amish kindred in Indiana, where the name of the disease came from. In fact, the name of "Amish brittle hair syndrome" is quite misleading; it suggests the disease exists only among the Amish. However, significant numbers of non-Amish persons have this disease, although it is rare in the general population.

Several other names have been used by the medical society for Amish brittle hair syndrome, such as nonphotosensitive trichothiodystrophy, hair-brain syndrome and BIDS syndrome. BIDS syndrome might be a name best describing the disease: **B**rittle hair, **I**ntellectual impairment, **D**ecreased fertility and **S**hort status. However, we have found that clinical manifestations significantly vary in our patients, some patients only have mild symptoms and many of the patients have normal status. We have also found that mood swings are quite common in these patients. Parents sometimes refer to their affected children as "moody".

Diagnosing this disease remains a challenge due to its rarity and the lack of knowledge of the disease in medical society. Most patients come to the DDC Clinic seeking an answer after they have been seen by dermatologists in their area. The disease, however, is quite common in the Amish community

in both Geauga and Holmes Counties. The DDC Clinic currently provides medical care for more than 10 children with this disease; most of them are from Holmes County.

The disease is poorly understood although some research breakthroughs have occurred over the last few years. The mutation of the gene TTDN1 is believed to be associated with the disease, although the exact mechanism of the disease remains unknown. We have found no information in medical literature regarding the treatment of the disease; therefore, continuous work through the DDC Clinic will be of great value for the affected families. We are encouraged by patients' hair responses following some topical treatments. Nutritional supplements and/or medications have improved the patients' moods and learning ability. Yet, much work still needs to be done in this direction.

Amish brittle hair syndrome is an autosomal recessive disorder, which means both parents are carriers but do not have the disease. Each child of carrier parents has a 25% chance of having the disease. Our clinic research laboratory has developed a DNA test for assessing the disease and carrier status. If you suspect relatives of having this disease, or would like to learn more, please do not hesitate to contact the DDC Clinic office at 440-632-1668.

The Sixth Annual Patchwork Benefit Evening – A Rousing Success

Friday, April 28th ~ a beautiful spring night for our Sixth Annual Patchwork Event. Due to the overwhelming successes of our previous Patchworks and the need for even more space, we found a new home in the elegant Signature of Solon WVI Country Club. Thanks to your generosity, the Patchwork Benefit netted the DDC Clinic for Special Needs Children \$66,583.89! Not only is this event much anticipated throughout the community, but it is essential for DDC, bringing in 9.43% of the total funds raised for this year.

An enormous thank you to our incredibly generous sponsors – each sponsor is critical to the success of Patchwork, as they jump start the event into the black before we even begin the evening. Our sponsors underwrote \$ 18,000.00 toward our grand total. Each year not only are we so thankful for our new friends, but so appreciative toward our faithful sponsors. We couldn't do it without you.

The evening began with wonderful hors d'oeuvres and music by Joe Marino and The Cleveland Dance Connection. As guests received their information folders, they were introduced to our theme of the night: "Did You Know...?" Throughout the evening, our guests were challenged to answer several questions highlighting some of the many achievements DDC has reached since our doors opened just 3 years ago. Just for participating, guests received a favor, and then had many opportunities to win prizes throughout the Live Auction just for being brave enough to answer a question. We had some very close calls and lots of fun, while further educating our guests about just some of the milestones and services DDC offers.

The highlight of the evening was the unveiling of the video filmed to honor the DDC Clinic in conjunction with the prestigious Crystal Award from the Johnson & Johnson Community Health Care Program. Dr. Wang and DDC received this award just days before Patchwork. Many guests and volunteers were moved to tears as we watched and heard



testimonies from the local and medical communities and most importantly families about all that DDC has done to change present and future lives. At the completion of the video, guests were moved to wonderful generosity by our gracious Celebrity Emcee Dee Perry from 90.3 FM WCPN National Public Radio and the always unpredictable and entertaining Professional Auctioneer Dave Rennolds for our Live Auction. Fantastic items

were created and lovingly handmade especially for Patchwork, such as benches, quilts, desks, tables, custom made furniture and clocks as well as unique packages including a



Personal Patterson Fun Fest, golf at the exclusive Sand Ridge Golf Course, a Royal Caribbean Cruise, OSU football tickets and passes to Disney as well as a basketball autographed by LeBron James. The wide array of items brought in record donations and fun.

Many other activities took place throughout the evening that allowed many to be winners as well. The silent auction with the *Auction Highlights* section offered more items than ever, and we continue to be astounded at the generosity, creativity and truly wonderful items donated by the community to support DDC. We had a mouth watering expanded bakery sale coordinated by our Amish moms; as well as instant winning opportunities in our *Fishin' for Fun and Treasure Chest*, where you are guaranteed to come away with at least as much as you put in. Our raffles were great successes once again, with the cash prize of \$500 and the Rolling Star Amish Quilt with additional prizes as well enticing a multitude. Even more excitement was generated during the Live Auction when the quilt winners donated the quilt into the Live Auction for even more bidding.

This fantastic evening with its wonderful success could not have happened without each of you. Our most sincere thanks go to Mark Kroner and the staff of Signature of Solon along with Mary Ruth Meyer for their untiring work to make this event and all the details possible and delightful. Thank you to our committee of volunteers and their families for making this happen – all your hours, sacrifices and patience were the key. Special thanks to Kathy McClure and staff of Proforma Lamar Printing; Patterson Fruit Farm; Joe Marino of the Cleveland Dance Connection; Cyndi Allen and her quilting friends; Cheryl Tausch of Design Wings and all of you, who donated, sponsored, attended and brought friends. Most of all, thank you to all the families, old and new friends of our children with special needs for making Patchwork more successful than ever.

Make plans now to join us once again at Signature of Solon for The Seventh Annual Patchwork Event ~ Saturday, March 24, 2007. See you there!

Upcoming Events

FEBRUARY 1, 2007

Holmes County Outreach Clinic, Holmes County Training Center

MARCH 1, 2007

Holmes County Outreach Clinic, Holmes County Training Center

MARCH 24, 2007

Seventh Annual Patchwork Benefit Auction and Dinner, Signature of Solon



6th Annual Amish Benefit Auction for DDC Clinic



The 6th Annual Amish Benefit Auction for DDC Clinic was held on November 11, 2006 at Middlefield Auction Barn. The Red Cross Bloodmobile, Cleveland Sight Center and DDC Clinic were all doing blood collection and screenings for those in attendance. The auction was well attended and raised approximately \$59,000.00 for DDC Clinic. "Everyone's efforts are applauded. The community continues to support DDC Clinic and for that we are very grateful", said Board President Darla Klein.