



# DDC Clinic Chronicle

THE NEWSLETTER OF DAS DEUTSCH CENTER FOR SPECIAL NEEDS CHILDREN

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## DDC Clinic Awarded Funding for New Love, Faith, and Family Project

Das Deutsch Center for Special Needs Children (DDC Clinic) is excited to announce that its new project *Love, Faith, and Family – Amish Genetic Disease Education and Care*, has been awarded \$820,000 in grants over the next four years. Local funding partners supporting the project include The Cleveland Foundation, Hershey Foundation, Michael Pender Memorial Fund, Susan and John Turben Foundation, Gordon Safran, and Elaine Motylinski and Roger Vaccariello. Up to \$410,000 will come from a matching grant from The Robert Wood Johnson Foundation (RWJF). This funding, to be used **exclusively** over the next four years for this new project, is dedicated to providing family outreach within the Amish community to promote comprehensive newborn screening for genetic and metabolic diseases. Through these grants, a greater number of children with genetic diseases will be diagnosed earlier and will receive specialized care at a younger age, allowing them to reach their full potential and greatly enhancing family life. Additionally, DDC Clinic plans to share these grant-funded services with Amish families outside of the Geauga County settlement.

DDC Clinic's mission is to enhance the quality of life for special needs children with metabolic and inherited disorders through early diagnosis and treatment, research, and education. The clinic is a unique collaboration of the Amish and non-Amish communities. Since its doors opened in May 2002, over 35 rare genetic disorders have been

identified in the community, often giving families answers they have been seeking.

Dr. Heng Wang, project director and DDC Clinic Medical Director, explains that the *Love, Faith, and Family* project is truly a team effort to build a healthier community. "The whole project is about LOVE and care. DDC Clinic's board truly desires to reach out to the families of the community to offer them help. FAITH emphasizes the clinic's commitment to respect the Amish religion and cultural traditions while FAMILY is the core value of the project," says Dr. Wang. According to Dr. Wang, the project has been supported by many community leaders and health professionals including the Bureau for Children with Medical Handicaps (BCMh), Middlefield Care Center, University Hospitals Health System, and the Cleveland Clinic Foundation. "DDC Clinic is looking forward to working with these supporters—and many other collaborators—to move the project forward," Dr. Wang states.

"As a fairly new and small not-for-profit organization," Dr. Tom Stone, DDC Clinic board president, commented, "we are thrilled that DDC Clinic's mission and vision have been recognized by such prestigious local and national foundations. It is exciting to hear that the clinic's program is considered a promising model by The Robert Wood Johnson Foundation. I believe that strong support from the community and local funders played a key role throughout the grant application process."

The Robert Wood Johnson Foundation matching grant was awarded under its Local Initiative Funding Partners (LIFP) program, one of the Foundation's most highly competitive annual grantmaking programs. Using rigorous criteria, the LIFP program identifies community-based projects that develop new approaches and collaborations to improve health and health care for society's most vulnerable people. In 2003, the LIFP office received 320 very strong applications for the 2004 awards and only 19 projects (six percent) were selected. The Robert Wood Johnson Foundation, based in Princeton, NJ, is the nation's largest philanthropy devoted exclusively to health and health care.

Anyone interested in more detailed information regarding this project or becoming a local funding partner, please call the DDC Clinic office at 440.632.1668. To learn more about other exciting projects DDC Clinic currently has underway, or to become a supporter of these projects, please call the clinic office.

### Wish List

- \$125,000 for Clinic expansion
- Parking area expansion
- Handicap-accessible van
- Salary for van driver
- Nitrogen tanks
- Equipment for gene-chip technology
- Storage shed

## Patchwork Event Exceeds Goals

A new site and a new set-up helped propel this year's fourth annual Patchwork benefit auction and dinner into the record books, netting almost \$75,500 to help children unable to help themselves.

Guests, numbering over 250, enjoyed this year's Patchwork event at Gilmour Academy in Gates Mills.

Featured auction items included handmade quilts, OSU football tickets, an oak hall seat, and many pieces of accent furniture. Other items included spa and restaurant packages, gift baskets and tickets to sporting events, theatre and orchestra. The live auction brought in almost \$40,000 itself with items such as the FunFest Catered Pig Roast, 8' x 12' children's schoolhouse, custom-made pieces of furniture, and cherry bookcase each being sold two to three times.

Ms. Eileen McShea, WKYC-TV weather forecaster, emceed the event and joined auctioneer Dave Rennolds as they took the highest bids during the live auction.

Heng Wang, M.D., Medical Director of DDC Clinic said, "Success at DDC Clinic is achieved through the support of corporate and individual donors, volunteers, and funding received from grants and foundations. DDC Clinic's results make a positive impact on children with special needs in Northeastern Ohio and worldwide."

Kudos and thanks to our generous donors and everyone involved in such a successful evening. See you next year!



Dave Rennolds and Eileen McShea auctioning off the next item up for bid at Patchwork 2004.

## President's Letter

Dear Friends,

It is county fair season in the green fields of Northeast Ohio. In a few days, thousands of exhibitors will display their best cookies, quilts and critters in the cool barns of the fairgrounds. Each year my family spends time at our local fair checking out the fancy rabbits and chickens, sampling the food, and trying to catch a fish in the pond at the back corner of the fair. It is a time for fun and celebration. It is a time to be thankful for all the riches we enjoy in this country.

At the clinic, new partnerships are being established with research organizations around the world. Families from near and far are finding care and support through our doors. We are always thankful that we are able to participate in this good work. Thank you for joining us.

Have a wonderful day.

Sincerely,

*Tom Stone*



... and a little child shall lead them.  
*(Isaiah 11:6)*

## Fellowship Designated

We are pleased to announce that Dr. Wang, DDC Clinic Medical Director, has been designated a Fellow of the American Academy of Pediatrics (AAP).

Fellows of the AAP are recognized by their colleagues and the Board of Directors as demonstrating excellence in

training leading to Board Certification as well as demonstrating high ethical and professional principals and conduct.

The AAP now has more than 57,000 members. Of these members, about 40,000 have earned the designation of Fellow (FAAP).

## Fourth Annual Amish Auction Scheduled

For those who have attended DDC Clinic's Amish Auction before, and for those who have yet to attend—get ready and mark your calendars now!

DDC Clinic's Fourth Annual Amish Auction is approaching fast. Come join us for an evening of great deals, fast-talking professional auctioneers, and the opportunity to bid on a huge variety of handcrafted items in our live and silent auctions held throughout the evening. Enjoy a hot meal followed by fabulous desserts and take home

delectable baked goods from our Amish bake sale. Take your chance on winning one of our unique raffle items while enjoying unmatched ambiance and good friends. Come see why DDC Clinic's annual Amish Auction is one of our most popular fundraising events.

Mark your calendar and plan on bringing your family and friends—spend a perfect evening in beautiful Amish Country and help support the children of our clinic at the same time.

- What:** Fourth Annual Amish Auction
- When:** November 19, 4:00 p.m.— 10:00 p.m.
- Where:** Middlefield Auction Barn
- For more information:**  
Call DDC Clinic at 440.632.1668

***Volunteers and auction items always welcome. Call to make arrangements.***

## Benefit Dinners and Events Planned for 2005

In 2005, DDC Clinic, along with University Hospitals Health Systems Geauga Regional Hospital, will offer our community supporters a new and unique opportunity.

Your ticket to this event entitles you to join us for dinner and activities on one of three special evenings. Each evening will begin with a reception and activities at the beautiful Red Maple Inn in Burton. From there, a bus ride through Amish country will offer a breathtaking view of the area. One local "feature" stop will be included. The tour will end at the home of one of our Amish friends where you will be served a mouth-watering, traditional Amish wedding dinner.

Thank you to this year's title sponsor—**University Hospitals Health System Geauga Regional Hospital** and to our other community sponsors that have graciously joined in to support DDC Clinic.

Sponsorship opportunities and volunteer positions are still available. To learn more, please contact the DDC Clinic office at 440.632.1668. **Watch for more details coming soon!**



**CHECK IT OUT!**



If you haven't visited the DDC Clinic website lately, you're missing out! Our website was recently overhauled thanks, in part, to help received from D.E.W. Consulting.

Our website now contains more information than ever. Visit our site for: a genetics primer, research updates, lists of the disorders found in our community and of the services we provide, along with details on upcoming events and much, much more!

**www.DDCClinic.org**

**We Have A Lot to Learn - In Memory of Mae**

**Heng Wang, M.D., PhD**

Again I find myself looking at the colored picture, “Juicy Red Apples.” Mae made it for me just two weeks before she passed away. The picture, of a smiling little girl holding a basketful of apples, always brings back memories of Mae.

I first saw Mae nearly two years ago. I was puzzled by her disease that included tremors for months after birth, and then a continuous tilting of her head to the left with low muscle tone. I had to admit that we didn’t have a diagnosis. For over a year, we worked with several professional teams trying to overcome many challenges in Mae’s care, particularly the not knowing of what she was suffering from.

On Mae’s third birthday in January, two test results came back to DDC Clinic from different laboratories. They confirmed her terrible diagnosis of “chicken breast disease”, a rare, lethal disease. I had suspected it several weeks before, but hoped that I was wrong.

As I set out for Mae’s house to break the news to the family, I was faced with one of the toughest house calls of my career. It was snowing heavily that day—the day after her birthday. It was quite late by the time I had finished with my last patient, returned phone calls, and managed to find their newly relocated home. The young parents, each sitting in a rocking chair and holding a child, had wondered if I would even make it that night. Mae was trying hard to stay awake, since she really wanted to show me the birthday gifts she received. As I looked at Mae’s smiling face and the birthday cards surrounding her

bed, I struggled to put the proper words into sentences. As a physician, I could not imagine anything worse than to have to tell her parents that Mae might die in six months, and that things we could do for her were very limited.

In the next several months, we frequently consulted Dr. Holmes Morton at The Clinic for Special Children in Lancaster, PA—they have treated almost all of the patients with chicken breast disease. Through Dr. Morton, we came to know Dr. J. P. Jin, probably the only research scientist in the world currently funded by NIH to study this disease. During many long conversations, we discussed Mae’s care, short-term and long-term, and what might be the potential treatment and hope for these children in the future. Ultimately, I arranged for Dr. Jin to meet Mae and her parents in the hospital. As he watched Mae struggle for breath in PICU, no words were needed, he knew what we all knew—that we had to work harder and faster!

After several hospital stays, Mae was getting weaker and weaker. Eventually, Mae was taken home to spend her last days with her family. Many house calls were made to keep her comfortable and many challenges were encountered in her care. Truly, she was a fighter—she came back talking and laughing after her blood PCO2 was over 90 in PICU; and nobody could believe that she safely kept a peripheral IV for nearly six weeks. Sweet and bright, strong and determined, Mae was always a joy for all of us, even on her last day.

Sitting with Mae’s parents and extended fam-

ily for several hours the day after the funeral, we shared our thoughts, our loss, and our grief. We spoke about the difficulties in Mae’s care, but much more, we recalled many joyful moments Mae had brought to us—her urge for school time no matter how sick she was, her pride in showing us the album a friend made for her after she knew Mae would soon leave this world... Looking through a window, I saw several young Amish children playing outside. Perhaps they were too young to truly understand life and death, and maybe they wondered why so many sobbing people passed through the doors, and, if Mae would ever come back to play with them again... When I left the house for the clinic, I wondered if we knew the value of life any better than those children. Very often, we overlook what those special children bring to us. Three years of life were too brief for Mae—for anybody, however, in many ways, she changed our lives—the things we were doing and the ways we were thinking. Dr. Morton said years ago, “These special children are not just interesting medical problems, subjects of grants, and research. Nor should they be called burdens to their families and communities. They are children who need our help and, if we allow them to, they will teach us compassion . . . they will teach us to love. If we come to know these children as we should, they will make us better scientists, better physicians, and thoughtful people.” Indeed, these children and their families have always inspired our work at DDC Clinic. They serve as a constant reminder for us—we have a lot to learn as physicians, scientists, and ordinary people.

*To protect patient confidentiality, Mae’s real name has not been used.*

**Footnote: Chicken Breast Disease**

Chicken breast disease is a relatively new disease in our community but it is quite common in the Lancaster County Amish community in Pennsylvania. By 2000, more than 70 children from 33 families with this disease had been found in Lancaster. Chicken breast disease, in medical terms, is called “Amish nemaline myopathy”. Genetically, it is autosomal recessive—both Mom and Dad are carriers, but have no signs of the disease. Each of their children has a 25% chance of being born with the disease.

Typically, children with chicken breast disease develop tremors (an involuntary shaking or trembling) at birth or within a few days of birth, particularly in the jaw or leg. The tremors subside over the first two to three months of life. Patients then develop progressive muscle weakness and atrophy (shrinking), along with progressive contractures of shoulders and hips. The most prominent feature of the disease is a severe deformity of the chest

with chest wall stiffness. The shape of the chest deformity looks, literally, like a “chicken breast”, hence the disease name. The deformity of the chest is generally irreversible, and affected children usually die from respiratory failure in the second year of life. The intelligence of these patients is usually normal.

Working with The Clinic for Special Children in Lancaster, Dr. Leslie Biesecker’s team from NIH has found that the disease is caused by a mutation of the slow skeletal muscle troponin T gene in chromosome 19. The mutation results in a premature stop codon at the place of amino acid 180. The research from Dr. J. P. Jin’s laboratory at Case Western has showed a complete loss of slow muscle troponin T in these patients. Interestingly, the mutation in our patient is exactly the same as the one in Lancaster patients. We found, when doing genealogy analysis in our patient, that we have to go back at least six to eight generations to see the Lancaster patients’ last names, such as Beiler, Fisher and King.

Therefore, we speculate that our community might have had more than one undiagnosed patient in the past.

Treating chicken breast disease is a challenge. Simple stretching exercises might be helpful for slowing the disease progression. Other potential treatments are currently in trial or discussion, including the drug induction of fetal troponin T isoform expressions and the stop codon suppression with aminoglycoside antibiotics. The latter has been recently studied in cystic fibrosis and Duchenne muscular dystrophy patients with some promising results. Further research in this area may apply to many other genetic diseases with the premature stop codon mutation in our community and around the world.

*If you might have relatives with this disease, or would like to learn more, please contact the DDC Clinic office at 440.632.1668.*