



CLINIC FOR SPECIAL CHILDREN NEWSLETTER

VOLUME I NUMBER 10

* LANCASTER COUNTY, PENNSYLVANIA *

DECEMBER 1995



GREETINGS TO EACH OF YOU
IN THIS SEASON AS HOPE IS SOFTLY REBORN
AND LIFE'S POSSIBILITIES ARE CELEBRATED
FROM ALL OF US
AT THE CLINIC FOR SPECIAL CHILDREN

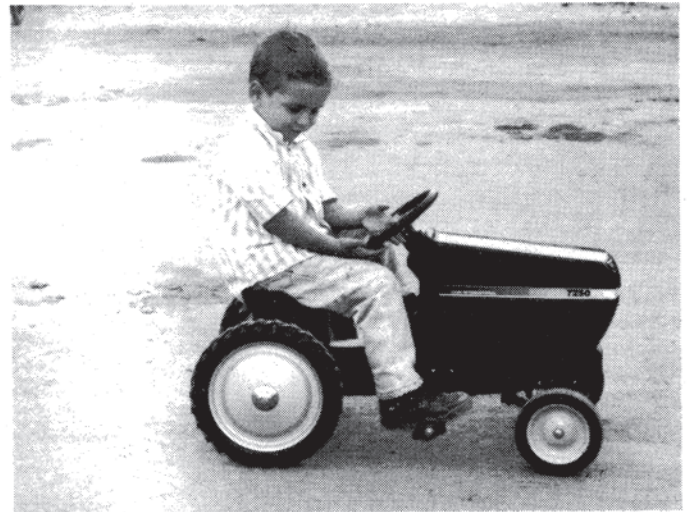


At this time of year we extend our thanks to all who help to make the work at the Clinic for Special Children possible and to the families and children who inspire it. Five years ago in November the beams of our building were raised and they stand sturdier than ever along with our determination to improve diagnosis and medical care for children with inherited metabolic disorders and other complex diseases.

A RECORD AUCTION

The fifth Annual Benefit Auction in September was the largest ever. The cool crisp fall air must have encouraged appetites and bidding as both exceeded our expectations. Seventy-eight beautiful large quilts in a variety of patterns including seven all white, sold on average for over \$500, record prices for our auction. Word of the best chicken barbecue in Lancaster County spread and by 3:00 p.m. the chicken was sold out as was one family's contribution of pig roast, and hundreds of pies, cookies, breads, and cakes baked by Amish and Mennonite women. Soft pretzels, fresh strawberry pies, and freshly baked donuts also disappeared by early afternoon. By 5:00 pm a chocolate bar, the only edible item in sight, was auctioned at a price that would excite the folks at Hershey. Next year we promise to have more food! Donated items auctioned included locally made furniture, an antique fainting couch, several Quaker style utility sheds, wood wagons, farm supplies, local crafts and dolls, hand crafted children's furniture, rockers, mid-wife's services, and much much more!

Several children at the auction were delighted to find new modes of transportation. Jordan Groff got his tractor! He seemed to know from the start that it was meant just for him. Two years ago during the auction, Jordan was critically ill with complications from maple syrup urine disease. This year, fully recovered and in good health, it was a joy to see him drive his tractor. The Mortons' three children managed to bid successfully on a pony while their father, Dr. Morton, was



admitting a sick child at the hospital and their mother was observing the quilt auction. We'd like to report the children and the pony are all doing well and enjoying each other (the parents have survived this demonstration of independence). Mary, Sarah, and Paul Morton chose to name their pony "Third Saturday of September" in honor of the Clinic's auction each year- a meaningful day to the Morton family. She is called "Saturday" for short. If you can't remember the auction day, just ask about the pony!

The auction's great success comes from the generosity of so many who contribute in one way or another. The lovely quilts come from the hearts of many women who know and love the special children who come to the Clinic. The crafts and furniture are made by hands with skill, but also hearts that care. As one visitor wrote us after the auction "I...see the unconditional love and support that so many people have for these children you all treat. I could really feel it sitting there in the auction." While Dr. Morton was detained elsewhere with an emergency call, there was a need to ask if there was a doctor "in the house". It was gratifying to see how many stood up who had joined us for the day - physicians from Lancaster,

Johns Hopkins, Kennedy Krieger Institute, Dupont, Hershey Medical Center, and Children's Hospital of Philadelphia. The auction serves not only as a time to buy a beautiful quilt or enjoy delicious food all for a worthy cause, but it also inspires many elsewhere in how a community of caring people work together. All of us at the clinic including the families who depend on us wish to express our thanks for this support. It offers us encouragement and the means to work even harder to improve care for children born with genetic disorders. It gives families with special children another reason to hope. It gives others ideas of how a community can work to accomplish so much. This year's total proceeds grew by 25% over last year's to provide support for the Clinic's operational budget. Without this vital support, we could not continue to operate with such a reasonable fee schedule for families who need us and continue the search for better ways to care for children with metabolic disorders and other difficult medical problems.

Thank you to the members of the Auction Committee who for the fifth year gave tirelessly of their time and energy to make the auction such a success: Mr. and Mrs. Leonard Hurst, Mr. and Mrs. Enos Hoover, Mr. and Mrs. Harvey Hoover, Mr. and Mrs. John Fisher, Mr. and Mrs. Steve Huyard, Mr. and Mrs. John Stoltzfus, Mr. and Mrs. Steve Beiler, Mr. and Mrs. Jacob Zook, Jr., Mr. and Mrs. Daniel Stoltzfus, Mr. and Mrs. Ernest Zimmerman, Mr. and Mrs. Daniel Lapp, Mr. Ralph Atkinson, and Miss Rebecca Huyard.



"Third Saturday of September"

SUPPORT IN NEW YORK

The auction was not the only fund raiser held this fall to benefit the Clinic. In Rochester, N Y, friends of the Colby family who bring their daughter to the Clinic, organized a pig roast fund raiser for the third year and the proceeds of nearly \$6000 were sent to support the Clinic's work to improve treatment for maple syrup urine disease. We express our thanks to all in the Rochester area for their support. There are now four MSUD patients in the central region of New York who receive their specialized care from the Clinic.

Note: Laura Colby is featured in the 1996 Mead Johnson calendar of special children for the month of July.

JOURNALS REQUESTED

The Clinic needs complete sets of old genetics journals to add to our reference library. Please call or write the clinic if you have issues available to verify what series we can use.

(717) 687-9407

GLUTARIC ACIDURIA CONFERENCE II

On December 7 our second scientific conference about glutaric aciduria was held at The Clinic. The first such meeting was in March of 1992. Most physicians and scientists who came to our first GA1 conference returned and we welcomed new participants from Dartmouth Medical Center, Children's Hospital in Philadelphia, Marburg Germany, Goteborg Sweden, Dublin Ireland, and Winnipeg, Canada. On December 8th physicians met with parents of children with glutaric aciduria. Families came from throughout the US as well as from Sweden & Germany. Amish families from Lancaster County provided a hot lunch for more than 90 participants.

The meeting with families was an important addition to the GA1 conference. There is no better way for physicians to appreciate the needs of children with glutaric aciduria than to meet children with the disorder who have been injured. There is no better way to understand the hope that research brings to parents, children, and doctors than to meet children with GA1 who have remained well because of advances in diagnosis & treatment. Equally important, some families who came to the conference had never met another family with a child affected by GA1. New friendships, advice about medical care, home care, education for the injured child, as well as parent to parent discussions about forming a parent support group to advocate screening, treatment, and research will serve many important lasting purposes.

In June of 1988 when the first Amish boy was recognized to have GA1, the disorder was thought to be rare and untreatable. It is now apparent that neither is true. As awareness of the disorder has grown within the medical community and as methods for diagnosis have become better and more widely available, GA1 has emerged as one of the more common metabolic disorders in the general population. Unrecognized cases may be children with unexplained *cerebral palsy*, children with large heads and low muscle tone who are slow to sit or walk, infants and children with subdural collections of fluid who may have life-threatening brain hemorrhages after minor falls and have too often been mistaken for abused children. Clusters of cases of GA1 have been recognized in Israel, Germany, Sweden, Ireland, among native Americans in the US and Canada, within all of the Amish subpopulations throughout Pennsylvania and one Amish settlement in northern New York state. Unrecognized and untreated GA1 has a very high morbidity & mortality. More than 90% of untreated cases will suffer major brain injuries and as many as 25% of untreated cases die before 5 years of age. Presentations at this conference leave no doubt that the disorder is treatable. **Collectively, brain injury has been prevented in more than 90% of cases that were diagnosed through testing of high-risk asymptomatic infants in Germany, Ireland, Sweden, and Pennsylvania.**

Remarkable progress in our understanding of the disease has been made between 1992 and 1995. Last year Dr. Stephen Goodman, University of Colorado, and his coworkers identified the Amish gene defect, and more than 10 others gene mutations, that cause the disorder. Carrier testing can now be

done at the Clinic to help identify high risk Amish couples and infants. Dr. Edwin Naylor and coworkers at Duke University have developed a filter paper based method for screening newborns for GA1. Dr. Naylor's supplemental screening program now tests 80,000 infants per year in Pennsylvania and provides a practical & economical way to reach high risk Amish populations outside of Lancaster County. Of note the most recent case of GA1 from Lancaster County was diagnosed through Dr. Naylor's program on the morning of the GA1 conference. Also, 3 of the 5 most recently diagnosed cases of GA1 in Pennsylvania were not Amish. In 1992, it was not known that the subdural fluid collections found in most infants with GA1 placed these infants at risk for subdural hemorrhages after minor head trauma. All physicians at the December 1995 conference knew of one or more cases that came to medical attention in this way. Such hemorrhages may precipitate major metabolic crisis and lead to basal ganglion injury. Tragically, in several cases the metabolic basis of the subdural effusions and brain injury was not recognized and parents were initially accused of child abuse. Child abuse teams are now aware of the need to routinely rule-out GA1 not only to prevent false accusations but also to allow appropriate treatment of the underlying metabolic disorder. We also now have a better understanding of the brain injury caused by GA1. In 1992, Drs. Morton & Kelley suggested that the risk of basal ganglion injury was highest between 4 and 18 months of age and by 6 years of age children with GA1 appear to be neurologically stable. This important observation has held true, none of the presenters at the 1995 conference knew of a patient who had experienced sudden basal ganglion injury after this age. In 1992 we also drew attention to the relevance of research by Dr. Micheal Johnston of Kennedy Krieger Institute and others about the *excitatory toxin mechanism of injury* to the basal ganglion. This model of glutamate receptor mediated injury now appears to explain the focus of the injury within selective regions of the brain and the age/maturation sensitive nature of the injury. More important, the model suggests new pharmacologic approaches to therapy that may limit the need for dietary therapy and hospitalizations in asymptomatic infants and may reverse the early stages of acute brain injury.

Progress has been impressive but much remains to be done. There has been little progress in our efforts to improve muscle control and speech for injured patients. Systematic studies of the movement disorder and therapeutic trials are badly needed. Gene based therapies await a better understanding of the relationship between gene, metabolism, and disease. Correction of biochemistry in the liver or in blood cell lines is unlikely to afford protection to the brain. Selective transplant of normal genes into nuclei of the basal ganglia may some day be possible but there is no reason to believe that such gene repair will reverse advanced brain injury. Successful gene therapy will ultimately depend upon our ability to recognize asymptomatic infants and protect them from injury. Education of health care providers about GA1 remains an important task. Many physicians, even at hospitals in Pennsylvania that serve high risk Amish populations, infrequently send appropriate biochemical tests on infants with findings suggestive of GA1 and do not support widespread screening. Until the standard of

general care improves for infants with GA1 and other metabolic disorders most infants with GA1 will only be diagnosed after severe brain injury has occurred.

Presenters, who collectively treat more than 150 children with GA1, included Dr. Holmes Morton from the Clinic for Special Children; Prof. Dr. Georg Hoffmann, Philipps Universitat Kinderklinik, Marburg, Germany; Dr. Martin Kyllerman, Department of Pediatrics, Goteborg University, Goteborg, Sweden; Dr. Eileen Naughten, Metabolic Department, Children's Hospital, Dublin, Ireland; and Dr. Cheryl Greenberg, Winnipeg, Manitoba. Other presenters who are involved in research related to GA1 included Drs. Richard I. Kelley and Michael Johnston from Kennedy Krieger Institute and Johns Hopkins in Baltimore; Dr. Stephen Goodman, University of Colorado; Drs. Steve Kahler and Johan Van Hove, Duke University; Dr. Charles Stanley, Children's Hospital of Philadelphia; Dr. Edwin Naylor of NEO GEN, Pittsburgh; Dr. Jim Filiano, Dartmouth Medical Center; and Dr. Amy Patterson of the FDA. Among other participants were physicians from Children's Hospitals of Philadelphia, Pittsburgh; Baylor Research Institute, Texas; Mass General in Boston and the FDA (Food and Drug Administration) in Washington, D.C.

MSUD GATHERING

The GA1 conference offered an opportunity to review treatment for maple syrup urine disease. Dr. Eileen Naughten in Ireland also has patients with MSUD and it was interesting for several specialists in attendance to compare notes with each other. On Saturday, December 9th, despite snow and ice Dr. Gerard Berry and Dr. Paul Thornton from Children's Hospital in Philadelphia, Dr. Vivian Shih from Massachusetts General in Boston, MA, Dr. Mike Gibson, Baylor, joined Drs. Morton, Naughten, Hoffmann, and Kyllerman for informal discussion about treatment for MSUD, particularly methods to manage critical illness and reverse cerebral edema. The session continued over dinner in the afternoon with about 25 MSUD families at the farm of Enos and Anna Mae Hoover.

NEW STAFF

We are happy to report the recent addition of two full time staff members at the Clinic. Lillie Rizack, R.N., is the new office pediatric nurse. A graduate of Franklin & Marshall College, she received her training from Lancaster General Hospital's School of Nursing and for the past few years worked with the pediatric oncology service at Hershey Medical Center, and also frequently volunteered at the clinic to help with our immunization program. Donna Robinson, R.N., C.P.N. joins our staff as a pediatric nurse practitioner. She was previously pediatric charge nurse at Lancaster General Hospital and cared for many of the clinic's patients in the hospital. A graduate of Eastern Mennonite College in Harrisonburg, VA, she recently completed her masters degree from the University of Pennsylvania. We are very fortunate to have Lillie and Donna on our staff. Both are talented, skilled professionals who demonstrate a sense of commitment to the work of the Clinic and the children and families it serves.

THE CLINIC FOR SPECIAL CHILDREN, INC.
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With new staff comes greater potential to fulfill our expectations of the scope of comprehensive care for children with metabolic disorders and other complex syndromes. A review of our current patient files over the last few months provided new staff with greater familiarity of our patients as well as an opportunity to examine each case to determine if we are doing everything we can for that child in terms of treatment plan, diet, needed referrals for adaptive equipment, therapy, etc. One area of need we addressed recently was related to psychological issues that affect families and children with chronic diseases. Counseling services are now available through the Clinic specifically suited to the needs of children and adolescents with genetic disorders who face unusual challenges in adapting and learning to live a meaningful and fulfilling life. Barbara Brown Wanta, M.S.N., R.N.C.S. of Wilmington, DE, joins us once a month as a specialized therapist in this work.

LECTURES GIVEN

The work at the Clinic continues to be of interest to a wider spectrum of pediatric specialists, medical students, and other hospitals. Although time away from clinical work even for short periods is difficult, Dr. Morton does give lectures on average of once a month regarding the importance of newborn screening and early recognition of a range of metabolic disorders and to discuss approaches to treatment. During the past year he lectured at hospitals in Allentown, Reading, Ephrata, York, Lewistown, and State College. He also gave Pediatric Grand Rounds at the University of Vermont Medical Center and Children's Hospital of Pittsburgh. Other presentations included the Annual Family Practice Review Program sponsored by Temple University and Lancaster General Hospital, the National Youth Science Camp held in WV, and the Child Neurology Society Meeting held in Baltimore in October.

Upcoming lectures in 1996 include a lecture about treatment of genetic diseases at the American Association for the Advancement of Science annual meeting in Baltimore in February, the keynote lecture for the Family Practice Review Program in March about the Special Child in General Practice, and a special memorial lecture to be given at Stanford University Medical Center in the spring.



*The Clinic for Special Children is a non-profit diagnostic and primary pediatric medical service for children with inherited metabolic disorders in Lancaster County, Pennsylvania. The clinic serves Old Order Amish, Mennonite and other families who suffer from a high incidence of genetic diseases such as glutaric aciduria and maple syrup urine disease. Clinic services include infant testing programs for early diagnosis, primary medical care to prevent devastating effects of metabolic diseases, clinical research to improve treatment, and services to support the needs of parents. The Clinic is funded through a combination of fees for services, benefit auction proceeds and private contributions. The Clinic does not seek or accept federal or state support and is tax exempt under IRS 501 (c)(3), ID # 23-2555373.
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