CLINIC FOR SPECIAL CHILDREN NEWSLETTER

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GREETINGS !
WE WISH THAT ALL CAN SHARE IN THE SPIRIT OF HOPE RENEWED BY THIS JOYOUS SEASON!

COMMUNITY SPIRIT AT WORK

BENEFIT AUCTION 1992

The second Annual Benefit Auction organized by parents for the clinic was a great success. On September 19th more than five thousand joined us for the day at the Leola Produce Auction shed in Lancaster County to enjoy the fun, food, and fellowship in support of the Clinic. Many at the auction commented on the strong evidence of community spirit. It was another day of inspiration for those of us at the Clinic. One respected Mennonite leader said he had never seen the combined efforts of the local Mennonite and Old Order Amish communities at work quite like this. We thank all those who came and who supported the Clinic through their purchases, donations, and time and effort. Your generous spirit is appreciated and is essential to the future of the Clinic. **THANK YOU!**

Fed by chicken barbecue, which some said was the best they had ever tasted, and freshly made subs, soft pretzels, ice cream, fresh strawberry and peach pies, and tables of baked goods, bidders got in the spirit to buy everything from exquisite handmade quilts to farm machinery to ponies, chickens and clover. Among items auctioned was a handmade set of oak bedroom furniture made for the occasion by an Amish father; an antique velvet crazy patch comforter made by the great-grandmother of two children cared for by the clinic; a new Zook built farm wagon with steel wheels; over 125 handmade quilts and wall hangings, many made specially for the sale by mothers, grandmothers, and aunts of children who are patients at the Clinic; locally made lawn furniture, kennels, and a wooden swing set; hickory rockers; hand made dolls, wagons, toys and collector's Winross trucks. A cake decorated to look like the clinic was auctioned and bought at least three times in a show of spirited bidding before its flavor was finally enjoyed.

The auctioneers donated their services for the day. With their enthusiasm and rapport with bidders in full swing, they contributed much to the spirit of the day.

The final tally for the sale after expenses was $95,288.58. With related donations and sale of greeting cards, the grand total is **$99,064.99 !** This is over $20,000 more than was raised at last

Photograph by Jim Stansbury

year's auction and all of us associated with the Clinic are extremely grateful for this support. The Clinic could not operate without it.

We thank members of the community who donated items to make this year's sale so successful. Every item helped! We especially want to thank the group of parents who gave of themselves so generously to organize the auction. They are so efficient and dedicated that within a week of this auction they were already talking about next year's. Special thanks to organizing committee members: 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. Ralph Atkinson, and Miss Rebecca Huyard. Your efforts have made a difference to the Clinic and to many special children. To the fellows who barbecued all that delicious chicken, you are also very appreciated!

A WORD ABOUT OUR FUNDING

The Clinic depends on three sources of funding for its operational support. Approximately 35% of our cost of operation is met through fees for services. Fees are kept as low as possible to encourage our model of preventive care for chronically ill children and participation in the infant testing program for glutaric aciduria. Very few of our families have insurance or state assistance and it is of critical importance to maintain an affordable fee schedule. To help maintain these low fees, parents organize an annual benefit auction to raise funds on a community wide basis to supplement the Clinic's income. In this way the local community of Amish and Mennonite families not only learn of the Clinic's services but participate in its support. The Auction this year will fund approximately another 35% of our projected cost of operation for the coming year. The balance of our funding needs are met through other contributions from individuals and foundations.

Our last fiscal year, October 1991 through September 1992, of our total operational expenses, 75.6 % was for direct program services; 18% was for administrative costs; and 6.4% was fund raising and development.

Funding from contributions and foundations also supports the very active and productive research and educational programs at the Clinic.

WE NEED AND WELCOME YOUR SUPPORT !

RESEARCH ENDOWMENT FUND STARTED

A grant from the Philip D. Reed Foundation, Inc. helped launch the Research Endowment Fund to provide long term support for the Clinic's laboratory research and clinical investigation. In addition to diagnostic and pediatric care for children with inherited disorders, the Clinic's mission also includes study of these disorders to understand and improve care and dissemination of this information so that it may help others. We are very grateful to the Philip D. Reed Foundation for its support and for sharing the Clinic's commitment to this effort.

While the Endowment Fund grows to a supporting level through its yield of interest, the Clinic will continue to rely on other donations to support the research program.

ADVANCES IN TREATMENT OF MSUD

Maple syrup urine disease is in many respects the prototype of the inherited disorders managed by the clinic. If diagnosed within a few days after birth and managed well then infants with MSUD grow and develop normally. Poor nutritional management and poorly treated episodes of metabolic crisis will result in brain injury with mental retardation, severe motor disabilities, or death at an early age. Outside of Lancaster County the disorder is very rare, currently we manage 26 Mennonite children who have MSUD which is approximately 1/4 of all children with the disorder in the United States. In 2 years we have had more than 200 outpatient visits and performed amino acid analysis on more than 300 blood samples from infants and children who have MSUD. In the past 30 months 10 Mennonite infants have been diagnosed in Pennsylvania, 7 were diagnosed at the Clinic, and 9 of the 10 new cases are routinely cared for by the Clinic. The Clinic is unique in the world for the number of patients with maple syrup urine disease treated, for the effectiveness of our management, and for our emphasis upon outpatient care.

The importance of our emphasis upon recognition of asymptomatic infants and early outpatient intervention in metabolic crisis is apparent. In Canada 9 neonates diagnosed through newborn

screening were in hospital for an average of 61 days after diagnosis. Of the 7 infants diagnosed at the Clinic, 5 were managed as outpatients at a cost of less than $500 each. The three Pennsylvania Mennonite infants who were diagnosed and initially treated by tertiary care centers in Philadelphia had combined bills for their initial hospitalizations in excess of $200,000. The two infants diagnosed by the Clinic who required hospitalization, whose degree of metabolic illness was similar to the Mennonite infants treated in Philadelphia, were managed by Dr. Morton at the Lancaster General Hospital. They were hospitalized for 5 and 6 days and the total cost of their care was less than $5000. In Canada the rate of rehospitalization of children with maple syrup urine disease after the neonatal period because of metabolic crisis averaged 19 days per patient per year of follow-up. None of the infants diagnosed and managed by the Clinic has been rehospitalized. Among the 16 patients older than 2 years of age, 6 have been hospitalized at Lancaster General for management of acute metabolic illness. The average stay was 1.5 days and there were a total of 12 hospital days. Our overall rate of rehospitalization is less than 1/4 day per patient per year. None of the patients managed by the Clinic have required admission to a tertiary care center because of an acute metabolic illness. Most important, the growth and development of our newly diagnosed children are normal. From this experience and model of care, we are developing similar approaches for other genetic disorders.

FROM UNDER A MIMOSA TREE

A mimosa tree in the backyard of an Amish home provided shade for discussion between Amish parents and doctors from the Clinic for Special Children, Kennedy Krieger Institute, and Johns Hopkins University in Baltimore, Maryland. The parents represented a dozen of at least 27 families who have lost children to a deteriorating genetic muscle disorder known locally as chicken breast disease because of the characteristic deformity of the chest in the later stages of illness. The syndrome is not described in medical literature and no effective treatment is available. Of the more than 60 children over the last 15 years who inherited the fatal disorder nearly all died by the age of two years. While doctors were gathering information from parents about their children's illness, a neurologist & his technician were conducting electrical tests of nerves and muscles on two newly diagnosed infants with equipment brought from Johns Hopkins and hooked up to a generator in the barn. Clues to understanding this deadly disorder are beginning to emerge and we hope someday to report this as a treatable disorder, but much is still unknown.

CONFERENCE HIGHLIGHTS PROGRESS ON INHERITED DISEASES

The Clinic for Special Children in collaboration with the Lancaster based Louise von Hess Foundation for Medical Education sponsored a conference on November 18, 1992, on "Inherited Diseases of the Amish and Mennonite People: From Research to Preventive Care". In the first lecture Dr. Holmes Morton, Clinic Director, reviewed changes during the last 20 years in diagnosis and treatment of Maple Syrup Urine Disease and more recent progress in early recognition and treatment of Glutaric Aciduria. Dr. Morton's second lecture gave an overview of 45 other inherited disorders found in the Amish and Mennonite populations of Pennsylvania. His lecture emphasized that more than half of these disorders are readily treatable. Early recognition and informed primary health care can prevent severe illness or disability. The final lecture of the evening was given by Dr. Victor A. McKusick, Emeritus Professor of Medical Genetics and former Physician-in-Chief at Johns Hopkins University School of Medicine, whose catalog of genetic diseases Mendelian Inheritance in Man is in its 10th edition and one of the most widely used genetic reference books in the world. Dr. McKusick spoke about genetic diseases of the Amish in part from an historic perspective. His studies of several types of dwarfism among the Amish in Lancaster are among the best known studies of population genetics in the medical literature. Dr. McKusick is also one of the founders of the the Human Genome project which is a joint project involving many of the major genetic research institutions in the United States in an effort to describe all the gene locations on the human chromosomes. His lecture also looked forward to the day when such information will provide new opportunities for treatment of genetic diseases.

The conference, which was directed to Lancaster area medical practitioners to increase awareness and information about inherited disorders, had excellent attendance from central Pennsylvania with others from Johns Hopkins in Baltimore and Yale University in Connecticut joining the meeting. The conference is part of the Clinic's effort to share with medical practitioners what is learned from our clinical experiences and research in an effort to stimulate interest in recognition and treatment of genetic disorders. Earlier this month a lecture about maple syrup urine disease was given at the Soldiers & Sailors Memorial Hospital in Penn Yan New York which serves a new settlement of 200 Mennonite families from Lancaster County. Over the next 6 months Dr. Morton has lectures scheduled for the Family Practice Review Course at Temple University in Philadelphia, the Pediatric Review Course in Charleston West Virginia, and for a Clinical Research Seminar at The Rockefeller University in New York.

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HELP IS ON THE WAY!

For three years Dr. Holmes Morton has worked alone to provide medical coverage 24 hours a day 7 days a week at the Clinic. All admissions to Lancaster General Hospital for management of metabolic illnesses and other complex illnesses related to genetic disorders have been directly under his care. All work in the Clinic's highly complex laboratory including calibration and maintenance of equipment and all emergency sample analysis for hospitalized children has been done by Dr. Morton. Although the original focus of the work at the Clinic was to be upon glutaric aciduria and maple syrup urine disease over the past three years more than 20 inherited disorders of biochemistry and 20 other genetic syndromes have been seen at the Clinic. For some of these disorders effective treatment is known but requires complex management such as that required to successfully treat maple syrup urine disease and glutaric aciduria. For most of these disorders medical care needs to be improved by further research and by development of comprehensive care plans. For still others little is known and current treatment is ineffective but compassionate care of the affected child and family is both difficult and important. The effectiveness of our approach to care is increasingly apparent to all who work at the Clinic and use our services but is equally apparent that if the Clinic is to endure, some of the day to day responsibilities for care must be shared with others.

The Clinic is very pleased to announce that Dr. Richard I. Kelley of Kennedy Krieger Institute and Johns Hopkins University will be joining the Clinic to provide clinical services one day a week. Dr. Kelley, a pediatrician, geneticist, and specialist in metabolic disorders, has been involved with the Clinic as a board member and has volunteered his time and expertise in many ways for the Clinic and its patients.

We also look forward to the possibility of a pediatric neurologist joining our medical staff on a part-time basis for the next several months.

The Clinic for Special Children is a non-profit diagnostic and primary medical service for children with inherited metabolic disorders in Lancaster County, Pennsylvania. The clinic serves Old Order Amish and Mennonite families who suffer from a high incidence of genetic diseases such as glutaric aciduria and maple syrup urine disease. Clinic services include an infant testing program for early diagnosis, primary medical care to prevent devastating effects of metabolic diseases during common childhood illnesses, clinical research to improve treatment, and services to support the needs of parents. The Clinic is funded through a combination of fees for services based on cost and private contributions. The Clinic does not seek or accept federal or state support.