CLINIC FOR SPECIAL CHILDREN NEWSLETTER

VOLUME I NUMBER 27 * LANCASTER COUNTY, PENNSYLVANIA * SUMMER 2008

2008 BENEFIT AUCTIONS

JULY 12 ~ SHILOH, OHIO

JULY 19 ~ SHIPPENSBURG, PA

SEPTEMBER 13 ~ BLAIR COUNTY, PA

SEPTEMBER 20 ~ LANCASTER COUNTY, PA

The Clinic for Special Children welcomes Dr. Nicholas Rider to our staff officially as of July 21, 2008. Many patients and families may have already met Dr. Rider during the past two years through his collaboration with the Clinic in research and treatment for children with immune deficiency syndromes and Cartilage Hair Hypoplasia dwarfism. All of us at the Clinic have enjoyed his presence here for short visits during the last two years and look forward to seeing him at the clinic as a full time member of our staff.

Dr Rider joins the Clinic from Hershey Medical Center where he completed his residency in internal medicine and pediatrics, serving as chief resident, and recently completed a two year fellowship in allergy and immunology. While at Hershey, Dr. Rider received the Arnold P. Gold Foundation Humanism and Excellence in Teaching Award in 2007 and 2006, the Pediatric Residency 4th Year Teaching Award, and the Mary Louise Witmer Jones Resident Humanitarian Award. Dr. Rider is board certified in pediatrics, internal medicine and will receive certification in allergy-immunology in the near future. From Williamsport, Pennsylvania, he is a graduate of Millersville University and the University of Health Sciences College of Osteopathic Medicine in Kansas City, Missouri.

Dr. Rider's training as an internist will help us meet the challenges of caring for our adult age patients with MSUD, GA1, and MCADD who are now over 18 years of age. Teenagers and adults with Cartilage Hair Hypoplasia are at high risk for opportunistic infection,autoimmune disorders, and malignancies. Adults with Pyruvate Kinase Deficiency have multiple complications and we increasingly encounter presentations of inherited problems in adults such as LMNA cardiomyopathy. His interest and research in immunodeficiency diseases will expand the Clinic's capability to diagnose and treat children with these disorders.

We also extend a special welcome to Nick's wife, Tonya, and their two year old son, Zachary, who already knows where all the toy trucks are parked in the Clinic's waiting room.

REFLECTIONS ON WORKING AT THE CLINIC....

from Dr. Rider

When doctors and scientists unite to solve clinical problems, the end result is improved patient care today and a lasting contribution to the health of future generations. This model was first demonstrated for me two years ago when I spent one month at the Clinic as an Internal Medicine-Pediatrics resident. At that time, my hope was that I would learn more about metabolic diseases; however, I came away with a changed view on disease prevention and treatment. I learned how caring for the community, family and individual by understanding population genetics and pathophysiology can lead to efficient and cost-effective patient care.

One month at the Clinic during residency changed my perspective on medical practice by showing me how 'translational' medicine can be practiced. Dr. Morton and Dr. Strauss provided mentorship and sparked an excitement about the way that they practice. We spent hours talking about clinical problems and thinking about how these illnesses may be diagnosed earlier, treated more effectively and understood more clearly. I enjoy this work tremendously and thoroughly enjoy working with the people at the Clinic.

I have been asked several times why I wanted to work at the Clinic rather than follow a more traditional route of entering subspecialty practice. Although it wasn't apparent to me initially, work at the Clinic is what I had always hoped for as a physician. While in college and considering a career in medicine, I wondered if a practice setting existed where I could talk to my patients about biochemistry and it's impact upon their health. During medical school, I remember hoping that I could make a contribution to medical knowledge through clinical research and patient care. Throughout fellowship, I have become interested in understanding the genetic basis of disease for the purpose of preventing illness. In hindsight, I have always hoped that I could practice at the Clinic for Special Children; until two years ago, I did not know that such an opportunity existed.

Since May of 2006, I have come to the Clinic on a weekly basis to study and work with Dr. Morton, Dr. Strauss, and Dr. Puffenberger. The time spent in Strasburg has been extremely fulfilling and meaningful to me. I am thrilled to have the opportunity to join the Clinic staff, and to have the privilege to serve the patients of this community.

GATHERING SUPPORT

Summer brings two benefit auctions in July and the anticipation for two more in September. Together, all four events help fund about 28% of the Clinic's annual operating costs. They also provide a time for families with special children to meet each other, for others to learn and be inspired through the lives of the children and their families. **Please join us at an auction. We need your support.**

Shiloh, Ohio ~ July 12 Families in Shiloh, Ohio, sponsored the fifth auction in that region, and we are very grateful for the support growing for the Clinic from this community.

Shippensburg, PA ~ July 19 In addition to 44 quilts, tops and wall hangings, a new carriage and furniture, this auction always has many garden items and beautiful plants donated for sale. Home made ice cream and lemonade disappeared quickly on a hot July day.

Blair County, PA ~ September 13 Beautiful quilts, hand crafted furniture, great chicken barbecue, soft pretzels, plants, especially mums for fall, invite bidders to Morrison's Cove Produce Auction in Central Pennsylvania for the 12th annual auction to benefit the clinic. Contact Amos at 814-793-3634, Eli at 814-793-3010 or Paul Ray at 814-224-5442 for information or donations for the auction.

Lancaster County, PA ~ September 20

The 18th annual auction for the Clinic in Lancaster County will start at 8:30 on September 20 at the Leola Produce Auction. For those who arrive early, breakfast including omlets and fresh donuts will be available. Dr. Morton, Dr. Strauss and Dr. Rider will offer a few remarks around 11:00AM. followed by the quilt sale and furniture auction block. Lawn furniture and Gift Certificates will be sold around 1:00PM.

Quilts will include a Postage Stamp, Broken Star, various applique and Center Diamond designs. Locally made cherry and oak furniture, garden and farm items, crafts, baked goods and as always, fresh subs, and the best chicken and pork barbecue.

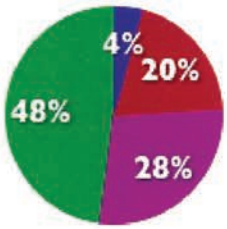
Call one of the following if you have items to donate or for more information: 717-626-4863; 717-354-5415; or 717-656-9694. The Leola Produce Auction is located on Brethren Church Road, north off Rt. #23 in Leola (between Lancaster and New Holland)

Thank you to all who volunteer; all who donate, all who come; and all who bid and buy. We thank the children who inspire all of us to work harder and to keep the Clinic growing and thriving.

WHY WE NEED SUPPORT

For the past fifteen years, the cost of a routine clinic visit has been $35. We have not raised this rate in spite of our rising costs. We want to make every effort to keep services at the Clinic affordable for all families, especially those who do not have insurance. The cost of running the Clinic and providing care has grown significantly and the income we receive from patient fees only covers about 20% of our total cost. The community sponsored auctions generally raise another 28% of the funds needed to meet the actual cost of providing medical services.
The balance needed each year to meet our expenses comes from donations from individuals and grants from foundations. All new equipment or special projects such as the Genetics of Hearing equipment, the new Light Cycler, research in the lab, clinical studies on GA I, new MSUD formula evaluation and many other projects designed to improve care are all funded through donations, special gifts, grants from foundations. A small portion of these costs are funded through a percentage of earnings from the Clinic's Research Endowment Fund. Our hope is that the Endowment Fund will grow large enough to cover a more significant percentage of our research, education and clinical studies costs. **We need major gifts from generous individuals to grow the Research Endowment Fund to meet our goal.**

PIECES OF THE PIE



4 % Research Endowment

20% Fees for Clinic Services

28% Auction Proceeds

48% Charitable Gifts

RAISING AWARENESS AND BUILDING SUPPORT

This spring lectures given by Dr. Morton combined with concerts by Cellist Matt Haimovitz and Paul Morton on classical guitar helped raise awareness and support for the Clinic. In March, Dr. Morton was invited to give the annual Rudene DiCarlo Memorial Lecture and resident rounds at Lucille Packard Children's Hospital in Palo Alto, CA. Following the lecture, a concert to celebrate the work of the Clinic was held at Stanford University's Cantor Museum. Many who attended the events donated to the Clinic's Research Endowment Fund including representatives from Affymetrix, Inc.

In April Dr. Morton gave the first of five CSC MacArthur Lectures at F&M College followed by a concert featuring Matt Haimovitz and Paul Morton. The event raised $25,000 from F&M for the Clinic's Research Endowment Fund. Individual donations from those who attended also contributed to the Fund.

A CHALLENGE TO MATCH

We have another call for a matching gift challenge. The Norman and Elizabeth Hahn Family Foundation will donate **$50,000** to the Clinic **IF** we can match this amount with other donations before December 1, 2008. **Please help us meet this goal.** We thank Mr. and Mrs. Hahn for this generous challenge gift and for their commitment of support for the Clinic.

IMPACT AWARD

The Clinic for Special Children was selected for a 2008 GlaxoSmith-Kline IMPACT Award. Recognized for its success and effectiveness in its overall innovative work and for it's significant impact in the community's access to healthcare, the Clinic was granted a gift of $40,000 to meet the needs of the organization. The Clinic was one of eight non-profit health care organizations in the Greater Philadelphia - Southeastern Pennsylvania Region to be selected for the award this year.
Funds received from this award were used to help replace and upgrade out dated computer equipment and software programs. This new technology will improve patient record information and support more effective access to information and ability to process data related to clinical and laboratory research. We are very grateful to GlaxoSmith-Kline for it's recognition of the Clinic and for the support granted to our organization.

Hearing Lab Almost Ready

With thanks from several donors including the Strasburg Area Sertoma Club, Dr. Morton's MacArthur grant, the Neuber Charitable Trust, the Genetics of Hearing Lab is almost complete and we will start scheduling hearing exams in early fall.

LABORATORY NOTES

from Erik Puffenberger, PhD., Lab Director

We continue to perform diagnostic and carrier testing for over 75 different mutations found in the Amish and Mennonite populations. Currently, we run over 375 carrier tests per year. Last summer, we added a new piece of equipment to the lab, a Roche LightCycler, provided by Franklin & Marshall College and Lancaster General Hospital. This system allows us to test for a specific mutation in a matter of hours. Our previous technique, PCR and DNA sequencing, was accurate, but not especially fast. The LightCycler will enable us to rapidly diagnose genetic disorders. It also reduces the amount of tech hours necessary as well as using fewer reagents. We are working with a company named TIB MolBiol to develop and validate an assay for each mutation. So far, we have over 30 functioning assays. We hope to double that number by the end of the year. Initial population screening using these assays with 100 random control DNA samples has shown that carrier frequencies for MSUD and Hirschsprung disease are quite high in the Mennonites, while carrier frequencies for Ellis-van Creveld syndrome and glutaric aciduria, type 1, are quite high in the Amish.

In the previous newsletter, we introduced **Eric Sherman**, an Ephrata High School student who has been spending several days a week at the clinic working on a variety of different research projects. Most notably, he has been studying glutaric aciduria, type 3 (GA3). This is a metabolic disorder similar to glutaric aciduria, type 1 (GA1), except that affected individuals remain healthy and never experience the devastating brain injury found in GA1.

While screening Old Order Amish infants and newborns for GA1 in the early 1990s, Dr. Morton identified three healthy children who excreted abnormal quantities of glutaric acid, but low or no 3-hydroxyglutaric acid, a pattern consistent with GA3. None of these three children harbored the known mutation for GA1 found in the Old Order Amish population. Eric initiated a genome-wide mapping study using Affymetrix 10K SNP Mapping Arrays and identified a large region on chromosome 7 common to all three GA3 children. This region contained 25 known genes, including *C7orf10*, a gene predicted to contain a mitochondrial targeting sequence and a coenzyme-A transferase domain. Eric sequenced this gene and found that all three Amish individuals were homozygous for a sequence variant (c.895C>T, Arg299Trp). He also sequenced an additional non-Amish GA3 patient who had visited the clinic years ago. That patient was found to carry the Amish variant and a second severe mutation, c.424C>T (Arg142Ter). Subsequently, Eric sequenced this gene from two additional GA3 patients (one Pakistani and one German) and found novel mutations in them as well. Future studies that determine the exact biochemical relationship between GA1 and GA3 may establish a foundation for more effective treatment of GA1.

LOOKING AHEAD

SUMMER CME COURSE

The Clinic will sponsor its second CME credit course for physicians on August 7th and 8th. The two day course titled *Diagnosis and Management of Genetic Disorders in Amish and Mennonite Communities* will provide pediatricians, family practitioners and other healthcare providers with an overview of the effect of inherited disorders upon the health of Amish and Mennonite children of Pennsylvania and discuss the impact of modern health care on the natural history of genetic disorders in these populations. Lectures will highlight the importance of Plain Communities in the history of medical genetics, review the molecular and biochemical diagnostic methods used, and present current understanding of population dynamics that cause high and low frequencies of specific inherited disorders in the Plain population and in other similar communities.

Topics presented by clinic staff and guest lecturers include:

Clinical Approach to the child with developmental delay,
Genomic approaches to diagnosis,
Treatment strategies in genomic medicine,
Relationship between gene and disease,
Physiology and genetics - an approach to hearing loss,
Genetics & the Radiologist and
Genetics & Pathology.

Please call the Clinic for information or to register (717) 687-9407.

FOURTH ANNUAL TRANSPLANT SYMPOSIUM ~ AUGUST 21

Children's Hospital of Pittsburgh UPMC and the Clinic for Special Children will jointly sponsor the ***Fourth Annual Children's Symposium on Transplantation for Maple Syrup Urine Disease*** on August 21 in Strasburg. The meeting will provide a forum for discussion of topics related to long term outcomes of transplant, neurodevelopmental and cognitive effects of transplant, research updates and questions for discussion from parents and patients. The transplant team from Pittsburgh and Clinic physicians will present the topics. To date, thirty one patients with MSUD and eight with Crigler Najjar Disease have received liver transplants at Children's Hospital of Pittsburgh.

F&M ~ CSC MacARTHUR LECTURE

The next lecture scheduled in the "MacArthur" lecture series sponsored by Franklin & Marshall College in collaboration with the Clinic for Special Children is scheduled for November 5, 2008 at 7:00 p.m.The lectures to be given by Kevin Strauss, M.D. and Erik Puffenberger, PhD. on translation of genetic research to clinical medicine will be open to the public and the F&M community. Their talk will be particularly aimed at helping Biology, Computer Science and Pre-Medical students understand the Clinic's concept of ***Bioinformatics*** as this term applies to human biology and medicine. The collaboration between the Clinic and F&M has led to a new major at F&M in Bioinformatics which is supported at the College by a prestigious grant from the Howard Hughes Medical Institute.

DR. MORTON INVITED TO LECTURE AT NIH

Dr. Morton will give an invitational *Director's Lecture,* A **Pediatrician's Perspective on the Human Genome Project** and the new area of medicine called Genomic Pediatrics at NIH on December 17, 2008. Dr. Francis Collins and Dr. Alan Guttmacher, the former Director and the current acting Director of the Human Genome Project issued the invitation to give the lecture.

The Clinic for Special Children was founded as a place where the knowledge from the fields of biochemistry and molecular genetics would be translated into new principles of medical care to help patients with inherited disorders. Although much of the knowledge of the human genome project was produced in laboratories far from the bedside, the translation of this information into medical practice ultimately requires a commitment to care for the patient. The every day practice of medicine is the next frontier of Translational Genetics. An understanding, an acceptance of the fact that many common illnesses arise in all people from genetic predispositions, but are nonetheless treatable, may ultimately be the most important contribution the Plain Communities and our Clinic for Special Children will make to the practice of medicine.

Leroy's Scarecrow

Diagnosis & Treatment of SCID CREATING A BONE MARROW DONOR DATA BASE

Severe combined immunodeficiency (SCID) is one of the most devastating forms of immune deficiency known. Babies with SCID are born without a part of immunity called the 'adaptive' immune system. The adaptive immune system has two major jobs: 1. Telling immune cells how to fight off and kill microbes (bacteria, viruses and fungi), and 2. Forming a memory of previous infections so that they can be prevented in the future. Genetic mutations causing SCID cripple the immune system and cause infants to become sick early in life. We have now cared for 8 SCID patients and know of 4 genetic mutations which cause SCID in our local community.

For most forms of SCID, bone marrow transplantation (BMT) is the only hope for a cure. Health following a BMT is strongly linked to two factors: age of the child at transplant and quality of the bone marrow match. Age is important because infections (especially viral infections) are more likely with time. Best results are with BMT at one to two months of age. Bone marrow matching, also called HLA (human leukocyte antigen) matching, determines how easily the bone marrow will grow and develop into immune cells (engraft) within the baby. We are currently working on ways to improve HLA matching and expedite transplantation for Amish and Mennonite babies.

In order to provide bone marrow to a baby born with SCID, a donor must first be identified. If the baby has a sibling with an identical HLA type, that sibling's bone marrow is used to treat SCID with a high probability of success. However, in many cases there is not a perfect sibling match and an unrelated donor's bone marrow is used. Instead of looking elsewhere, we believe that there will be a higher likelihood of finding an identical HLA match within the Plain Community for a baby affected with SCID. To determine this, we are planning a 'bone marrow drive' at the Clinic for Special Children in the coming year. Families considering participation in the bone marrow drive or wanting more information should contact the Clinic to let us know of your interest.

At the marrow drive, potential donors can have their HLA type determined from a small blood sample or swab of cheek cells. We will use this information to create a database of HLA types. When a baby is diagnosed with SCID, we will quickly search the database for a match. Our hope is that the HLA database will allow for more rapid and effective bone marrow transplantation for Amish and Mennonite children affected with SCID.

NEED FOR ULTRASOUND

We use ultrasound imaging for routine management of 26 genetic conditions (see Table attached). However, the cost of ultrasound exams is prohibitive for most of the families we serve who are primarily Old Order Amish and Mennonite, self pay and do not participate in insurance or medical assistance programs. About three years ago, we began to provide ultrasound studies free of charge using a Phillips HDI 5000 system loaned to us by the University of Maryland Amish Research Clinic. Collaborating cardiologists and radiologists from Children's Hospital of Philadelphia and Lancaster General Hospital donated professional time to analyze these studies. The capability for on-site ultrasound imaging improved our clinical follow-up, treatment compliance, and overall level of pediatric care. Moreover, it gave us a powerful way to investigate the physiology and disease progression of several genetic heart conditions that affect the Plain population.

We subsequently developed two IRB-approved clinical research protocols: one to study disease progression and response to treatment in hereditary hypertrophic and dilated cardiomyopathies, and another to determine patterns of middle cerebral artery blood velocity that precede stroke-like brain degeneration in children with glutaryl-CoA dehydrogenase deficiency. We also used ultrasound to diagnose and manage children with congenital heart disease. Since these studies began in 2005, we have collected cardiac or cerebrovascular ultrasound data from over 500 children and adults; we estimate this has saved the community well over $500,000 in medical costs.

The following offers one powerful example of how we use ultrasound to integrate genetic research and clinical care. We recently identified three large related Amish families with several members who developed relatively abrupt signs of congestive heart failure in early-mid adulthood and then died within 3 years. The most recently affected patient was a 33 year-old woman who died of sudden onset heart failure during a pregnancy last summer. Dr. Kevin Strauss visited these families at home, collected DNA, and within a week, Dr. Erik Puffenberger identified a dominant pathogenic mutation in LMNA (lamin A/C, 568C>T). Based on these findings we identified 20 young adults (children of affected individuals) who were at risk for the mutation, and invited them to the clinic for mutation testing, EKG, echocardiogram, and physical examination. Two local cardiologists volunteered their services for a day and all testing was done free of charge. We identified 7 presymptomatic mutation carriers, and could compare their EKG and ultrasound parameters to those of 13 unaffected siblings. We accomplished all of this work in one day at minimal cost; this type of study would have been complex and very expensive to execute at a university hospital.

Table of genetic conditions that benefit from ultrasound technology

ORGAN SYSTEM AND DISEASE	GENE	MUTATION	CLINICAL MANIFESTATION
HEART			
Trisomy 21	Ch. 21	Triploid copy #	Structural heart disease
Ellis-van Creveld syndrome	EVC	IVS13+5G>T	Structural heart disease
Bardet-Biedl syndrome	BBS1	1169T>G	Structural heart disease
Propionyl-CoA carboxylase deficiency	PCCB	1606A>G	Progressive dilated cardiomyopathy
Hypertrophic cardiomyopathy and lactic acidosis	SLC25A4	523delC	Progressive obstructive hypertrophic cardiomyopathy
Familial dilated cardiomyopathy	LMNA	568C>T	Adult-onset arrhythmias, DCM, and heart failure
Idiopathic infantile-onset hypertrophic cardiomyopathy	MYBPC3	IVS30+2T>G	Lethal infantile hypertrophic cardiomyopathy
Sitosterolemia	ABCG8	1720G>A	Accelerated coronary artery disease/atherosclerosis
Primary ciliary dyskinesia (Kartagener syndrome)	DNAH5	4346C>T	Dextrocardia/Situs inversus; complex structural disease
Marfan syndrome, infantile onset	-	-	Aortic root dilation/insufficiency
Ehlers-Danlos syndrome	-	-	Aortic root dilation/insufficiency
Congenital junctional ectopic tachycardia	-	-	Infantile-onset junctional ectopic tachycardia
Timothy syndrome	-	-	Syndromic long QTc syndrome
KIDNEY			
LYK5 deletion syndrome (PMSE syndrome)	LYK5	Tab deletion	Nephrocalcinosis with diabetes insipidus
McKusick-Kaufman syndrome	MKKS	250C>T	Congenital renal malformations
Cystinuria	SLC3A1	IVS6+2T>C	Nephrolithiasis
Wilm's tumor	WT1	Deletion	Renal tumor
Polycystic kidney disease, autosomal dominant	-	-	Cystic dysplasia
Congenital zneplasia/Potters syndrome	-	-	Lethal zneplasia and pulmonary hypoplasia
Prune belly syndrome	-	-	Hydroureter, hydronephrosis; various malformations
CEREBRAL VASCULATURE			
Glutaryl-CoA dehydrogenase deficiency	GCDH	1262C>T	Acute striatal necrosis; deep lenticulostriate distribution
Propionyl-CoA carboxylase deficiency	PCCB	1606A>G	Acute striatal necrosis; deep lenticulostriate distribution
LIVER, GALL BLADDER, SPLEEN			
Alpha-1 antitrypsin deficiency	SERPINA1	1096G>A	Progressive cirrhosis
Familial ivernhepatic cholestasis	ATP8B1	923G>T	Ivrnhepatic cholestasis, early-onset cirrhosis
Crigler-Najjar syndrome	UGT1A1	222C>A	Cholelithiasis/cholecystitis
Branched chain ketoacid dehydrogenase deficiency	BCKDHA	1312T>A	Cholelithiasis/cholecystitis
Pyruvate kinase deficiency	PKLR	1436G>A	Hemolytic anemia and splenomegaly

In LMNA disease, as with several other conditions listed in the table attached, we are now able to determine an individual's genetic risk before they get sick. This provides the opportunity to study the earliest structural and functional manifestations of a genetic disease and develop interventions that slow or prevent the progression to organ failure. It is a model of genomic medicine—the application of genetics to the everyday practice of medicine in an effort to solve difficult problems that face individuals and communities. To do this properly for cardiac conditions requires that we follow presymptomatic mutation carriers with serial ultrasound exams, and use this information to guide preventative care.

Unfortunately, the University of Maryland research group that loaned us the instrument had to reclaim it for another study and we can no longer offer this service. We are currently seeking support to purchase a new ultrasound system to continue as well as the funding needed to develop and provide more preventative, cost effective measures that address health care needs related to genetic diseases.

A NEW FORMULA FOR MSD

The Clinic for Special Children and Rick Finkel of Applied Nutrition have been working for the past three years to develop an improved formula mix for patients with Maple Syrup Disease. The formula is essential to the diet of MSD patients who are severely restricted in their intake of certain amino acids. The new formula, *Complex Essential MSD Drink Mix,* is based on many years of clinical study at CSC and designed to provide a more complete mineral and vitamin content with the essential amino and fatty acids needed for normal growth and development for MSD patients. Meetings with the FDA regarding the new infant version of the formula for MSD are scheduled for August. A similar study of a new infant formula for GA1 is in progress. The new formulas are another example of on-going research at the Clinic directly related to improving care for patients.

SUMMERTIME AT THE CLINIC

This spring and summer the Clinic has engaged many students in research projects and clinical studies. Dr. Morton, Dr. Strauss and Dr. Puffenberger taught a seminar class in medical genetics at Franklin & Marshall College for the spring semester. In May, **Mark Cathey**, a senior in medical school at the University of Virginia in Charlottesville, participated in a pediatric clinical elective rotation at the clinic. Mark plans to train in pediatrics after finishing medical school next spring.

Joan Brumbaugh, a 2008 graduate from Franklin and Marshall College, is working with Glutaric Aciduria type I this summer during a 10-week fellowship funded by the Eyler grant through F&M. Specifically, she is working with Drs. Morton and Strauss to investigate the biochemistry of the disease by looking at the effect of dietary amino acids. Through analysis of diet, development and other data of GA1 patients at the clinic, the goal is to quantify and summarize what we do and don't know about the disease to provide insights to treatment. Joan will begin medical school in August at George Washington University in Washington, D.C.

Shoshana Tell, a rising junior at Harvard, is working to develop a bone marrow database for the Amish and Mennonites of Lancaster County this summer during a 10-week fellowship funded by the Herchel Smith Undergraduate Research Fellowship at Harvard. Shoshana is working to develop this database with Dr. Puffenberger, due to the significant number of newborns born with Severe Combined Immune Deficiency (SCID), who require a bone marrow transplant as soon as possible. Matching a bone marrow donor with a recipient consists of determining that both have very similar Human Leukocyte Antigen (HLA) types, meaning that they have similar proteins displayed on the surface of cells. Hospital laboratories often use protein-based methods and charge upwards of $50,000 to find a match. The goal of this project is to thus develop a DNA-based HLA matching method that can be conducted in the Clinic's own labs and will be much more cost-efficient. Shoshana would like to thank Dr. Puffenberger, Dr. Morton, and Dr. Strauss for all of their help throughout the summer.

A SPECIAL AWARD FOR AN EXCEPTIONAL STUDENT

The Clinic is pleased to announce that **Eric Sherman** is the first recipient of a scholarship award designated by Dr. Morton in memory of his parents, Paul and Mary Morton. The award supports the study of a young student who shows unusual interest in the Clinic's work. The award recognizes extraordinary scientific curiosity in a student who demonstrates exceptional ability and promise as a future medical scientist. Eric, the son of Donna and Don Sherman of Ephrata and a junior at Ephrata High School, demonstrated extraordinary talent and ability in his research projects at the Clinic. The Morton Award is funded through a gift to the Endowment Fund from Dr. Morton's father, Paul Moton, and matched by a gift from Dr. Morton's MacArthur Award.

THANKS to Sue Sabado for many hours of volunteering at the Clinic on her days off as a nurse at LGH. She put together scrapbooks of the Clinic's history through newspaper clippings and magazine articles accumulated over the last 19 years. Great job, Sue, with our thanks!

CALLING FOR MORE BEANIE BABIES !

For the past few years Clinic staff have enjoyed dispensing donated beanie babies to children at the end of their clinic visits. It is a small reward for what most of them have to endure. Our beanie box is now empty! We have given away hundreds of beanie babies and need more. If any one has extra (clean!) beanie babies to donate, please send or call the clinic at 717-687-9407. We thank all, (and there were many, especially Marty and Scott Schoonover who started it all) who donated beanies in the past. Our patients absolutely love them and keep looking in the box for more!

NEW STAFF AT THE CLINIC

In addition to Dr. Rider, the Clinic also welcomes **Cris Mitchell** to our staff as a part time lab technician. Cris performs most of the Clinic's amino acid analyses and assists Dr. Puffenberger with other aspects of running the lab. She joined our lab in May, replacing **Steve Loscalzo**, a recent graduate of F&M who worked in the lab from January to May and participated in a research project with the Clinic ~ F&M collaboration. We wish Steve well in his graduate school plans.

DR. VICTOR A. McKUSICK

All of us at the Clinic will sadly miss Dr. Victor A. McKusick who died on July 23, 2008. Without his extraordinary contribution to the field of Genetics our work would not be possible. His early research, based at Johns Hopkins University, began here with the Amish population in Lancaster County. Many families we work with remember his early visits. His visit with us last summer was a memory we will cherish.

OUR WISH LIST

PLEASE consider a donation to the Clinic for Special Children. We need support more than ever for the following:

General support for patient services
Matching funds challenge
Funding for ultrasound equipment and services
SCID clinical and diagnostic studies and bone marrow donor project
Genetics of hearing loss project and equipment
Support for education and clinical Studies
Major gifts to grow the Clinic's Research and Endowment Fund

Clinic for Special Children
P.O. Box 128
Strasburg, Pennsylvania 17579

MISSION

The Clinic for Special Children was established in 1989 is a non-profit medical service for Amish and Mennonite children with genetic disorders. The Clinic serves children by translating advances in genetics into timely diagnoses,and accessible, comprehensive medical care, and by developing better understanding of heritable diseases.

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The Clinic for Special Children is a non-profit 501(c)(3) tax exempt organization and a registered charitable organization in Pennsylvania. Tax ID# 23-2555373

PA law requires us to advise that a copy of our official registration and financial information may be obtained from the PA Dept. of State by calling toll free 1-800-732-0999. Registration does not imply endorsement