Clinic *for* Special Children NEWSLETTER

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We are introducing our new logo - a combination of traditional symbols from medicine and from the communities served by the Clinic for Special Children. The forward facing notch conveys our sense of progress and determination in diagnosis and treatment of rare genetic disorders in children

FROM ALL OF US AT
THE CLINIC FOR SPECIAL CHILDREN:

Now in our 23rd year, we reflect on this year with great respect for our community and friends. As with so many non-profit organizations the Clinic was faced with impending financial hardship last year. We shared this concern with the community we serve, deacons, bishops, and church elders of both the Amish and Mennonite congregations during a special meeting last summer. We presented our needs to the community, and without hesitation, our friends responded with an outpouring of support. ***We thank you for this confident affirmation of our work on behalf of special children.*** *With this assurance we believe the Clinic will continue to serve special children for many years to come.*

We've come a long way in 23 years; over 125 unique disorders, 2,187 current patients served, and an estimated $23M in aggregate savings per year for the Plain Communities – all on an operating budget of $1.7M per year. Our model for medical care is an important beacon to the larger healthcare community and we depend on your support to keep costs low for families most in need. We are grateful and always mindful of the continued support of the Clinic. Ultimate value however, can only be measured in the health of the children who know the Clinic as their Medical Home.

WE THANK YOU

2012 BENEFIT AUCTIONS

THREE GONE:
June 2, Union County, PA
July 14, Shiloh, Ohio
July 21, Shippensburg, PA

TWO TO GO:
September 8, Blair County, PA
September 15, Lancaster County, PA

Auction Season is in full swing with a new auction event added this year. Families in **Union County** organized the first benefit auction for the Clinic in that region of Pennsylvania. On June 2nd the weather was bright, the turn out was greater than expected and the bidding went high for over 62 quilts, a kayak and other sporting goods, Bernina sewing equipment, clocks, furniture, hand crafts and many other items at the Buffalo Valley Produce Auction in Mifflinburg. Many Mennonite and Amish families from that region are connected to the Clinic through their own children or through relatives and friends.

We thank all who came out to support the Clinic and the families in the Mifflinburg region whose children have special medical needs. The success of this auction is a significant help for the Clinic.

On July 14th the Morton's travelled to **Shiloh, Ohio** to thank families in Ohio for their support of the Clinic for Special Children. This auction grows bigger every year and also supports the DDC in Middlefield, OH and will help build support for a new clinic serving Amish families in Holmes County. Hand made quilts, furniture, crafts, farm supplies, garden items, and of course delicious food all contributed to a very successful sale. Thank you to the Newswangers, Sauders, Burkholders, Martins and many other families who helped with this auction. This support is very much appreciated by all three clinics.

Dr. Morton and Dr. Strauss help sell a carriage in Shippensburg.

Dr. Strauss with baby Kendra

members to find a possible match for bone marrow transplant. Dr. Strauss made the two hour trip to the family's home to collect blood samples for typing by Dr. Puffenberger and Adam Heaps in the Clinic's lab. The baby's sister was a perfect match and the clinic forwarded the results to Dr. Bunin at CHOP to arrange for the bone marrow transplant. At two weeks of age the baby, her sister and parents travelled to CHOP for the transplant. Marrow was harvested from a very proud, brave sister and infused into the baby within a few hours. The family returned to their home the same day without having to stay one night at the hospital. This transplant may be one of the earliest ever performed and performed successfully on an outpatient basis. The Clinic sees the baby every few weeks to monitor progress.....and the progress is amazing! Now, three months old, she is healthy and learning patty cake from her 4 year brother.

UPDATES: PROGRESS IN 2011 RESEARCH AND CLINICAL CARE

Propionic acidemia: Prototype product (Applied Nutrition) soon ready for clinical trial.

Glutaric aciduria: Glutarade Jr. formula for GA1 developed by the clinic and Applied Nutrition: demand exceeding supply nationwide.

Cardiology: The Clinic is collaborating with Dr. Devyani Chowdhury, LGH Pediatric Cardiologist, in care and assessment of patients with cardiac complications of genetic syndromes such as ANT1/LMNA cardiomyopathies, Downs, and Ellis Van Crevelt dwarfism syndromes.

MSUD 'Cook-off' workshop and seminars held for teens with Maple Syrup Disease to teach special diet planning and cooking skills.

EVC Research: Clinic day held in August with Dr. Dennis Weiner, pediatric orthopedic surgeon, Akron, Ohio Children's Hospital.

Hearing Loss: Dr. Rob O'Reilly, DuPont helps the Clinic manage patients with complex hearing problems and collaborates with ongoing studies of the genetics of deafness to asses who might benefit from conventional hearing aids, bone conduction amplifiers, or cochlear implants.

Pretzel syndrome: The Weaver family sponsored a picnic in September attended by CHOP researchers who are studying a therapy that might ultimately be given to pregnant mothers.

Congenital Adrenal Hyperplasia: Family Day sponsored at the Clinic to review research and approaches to care.

Bipolar/Depression: The Clinic is collaborating with Dr. Sander Markx, Columbia University, to identify genetic risks for bipolar illness and major depression in Amish and Mennonite families.

CASPR2 Research: Collaboration continues with team at Columbia University, for potential new therapies for newborns.

Clinic's Genetics Laboratory: Renovations were completed in September and the Ion Torrent equipment for exome sequencing was installed in October. High- throughput sequencing for diagnostic purposes for such disorders as Yoder Dystonia will begin soon after the first of the year. We thank **Franklin & Marshall College** and **Lancaster General Health** for their support in expanding our laboratory's potential with the use of cutting edge diagnostic technology.

CLINIC OUTREACH AND EDUCATION

Pennsylania: Dr. Greg Mock opened a family practice in Belleville to help meet medical needs in Belleville/Bedford/ Somerset communities.

Dr. Holmes Morton and Paul Morton continue to help Dr. Mock and local community leaders in their effort to establish a new central PA Clinic for Special Needs.

Ohio: Dr. Olivia Wenger visited CSC in October and is moving forward with her plan to develop more consistent clinical services for special children in her community near Akron, Ohio.

Franklin and Marshall College Partnership: On Oct 19th we met with **Dr. Daniel Porterfield**, new president of F&M, to explore expanding the collaboration between the college and the Clinic that could include the new Public Health department at F&M. The Clinic is interested in working with students and faculty to develop support/education materials for parents and to help assess medical needs in central PA Plain communities.

Starting off in 2012:

Meeting with Dr. Alex Levin, Wills Eye Institute, January 30, to examine patients with GA1 of all ages for studies to understand complications related to retinal hemorrhages after minor head injuries and chronic headaches.

MSUD Diet Workshop with Alana Duffy, Applied Nutrition- January 17, 2012.

Maternal PKU Workshop with Dr. Berlin & PKU Follow-up Team: A Meeting with Teenagers and Young Adult Women with PKU. "What do you need to know about diet and protection of a baby from PKU during Pregnancy?" - January 18, 2012. Call 717-687-9407 to attend.

Properdin deficiency (hereditary meningitis) Family Day - January 26.

Congenital adrenal hyperplasia - Family Day -February/ March.

Treatment of Propioinic Acidemia- February 2012: Discussion of 2 year trial of a new formula to protect the heart, brain and prevent metabolic crisis.

Midwife CME course - Spring 2012.

A Review of the Genetic Causes of Stillbirth & Asphyxia. Newborn Resuscitation and the use of Hypothermia in the care of the Asphyxiated Newborn. Date to be determined.

Articles in press:

American Journal of Public Health, *One Community's Effort to Control Genetic Disease*, by Strauss, Puffenberger, Morton.

Nature, Feature article about the Clinic in upcoming February 2012 issue by Trisha Gura.

Public Library of Science One (PLoS One): "Genetic Mapping and Exome Sequencing Identify Variants Associated with Five Novel Diseases" Erik G. Puffenberger, Robert N. Jinks, et. al.....Nicholas L. Rider , Stacey Gabriel , D. Holmes Morton , Kevin A. Strauss [Co-Authored with Rob Jinks, 13 F&M students, and Eric Sherman].

Papers published in 2011:

Liver transplantation for classical maple syrup urine disease: long-term follow-up in 37 patients and comparative United network for organ sharing experience. J Pediatr. 2012 Jan;160(1):116-121. Mazariegos GV, Morton DH, Sindhi R, Soltys K, Nayyar N, Bond G, Shellmer D, Shneider B, Vockley J, Strauss KA.

Safety, efficacy and physiological actions of a lysine-free, arginine-rich formula to treat glutaryl-CoA dehydrogenase deficiency: focus on cerebral amino acid influx. MolGenet Metab. 2011 Sep-Oct;104(1-2):93-106. Strauss KA, Brumbaugh J, Duffy A, Wardley B, Robinson D, Hendrickson C, Tortorelli S, Moser AB, Puffenberger EG, Rider NL, Morton DH.

Erythrocyte pyruvate kinase deficiency in an old-order Amish cohort: longitudinal risk and disease management. Am J Hematol. 2011 Oct;86(10):827-34. Rider NL, Strauss KA, Brown K, Finkenstedt A, Puffenberger EG, Hendrickson CL, Robinson DL, Muenke N, Tselepis C, Saunders L, Zoller H, Morton DH.

Reduced thymic output, cell cycle abnormalities, and increased apoptosis of T lymphocytes in patients with cartilage-hair hypoplasia. J Allergy Clin Immunol. 2011 Jul;128(1):139-46. de la Fuente MA, Recher M, Rider NL, Strauss KA, Morton DH, Adair M, Bonilla FA, Ochs HD, Gelfand EW, Pessach IM, Walter JE, King A, Giliani S, Pai SY, Notarangelo LD.

Investigations of caspr2, an autoantigen of encephalitis and neuromyotonia. Ann Neurol. 2011 Feb;69(2):303-11.
Lancaster E, Huijbers MG, Bar V, Boronat A, Wong A, Martinez-Hernandez E, Wilson C, Jacobs D, Lai M, Walker RW, Graus F, Bataller L, Illa I, Markx S, Strauss KA, Peles E, Scherer SS, Dalmau J.

A novel mutation of LAMB2 in a multigenerational mennonite family reveals a new phenotypic variant of Pierson syndrome. Ophthalmology. 2011 Jun;118(6):1137-44.
Mohney BG, Pulido JS, Lindor NM, Hogan MC, Consugar MB, Peters J, Pankratz VS, Nasr SH, Smith SJ, Gloor J, Kubly V, Spencer D, Nielson R, Puffenberger EG, Strauss KA, Morton DH, Eldahdah L, Harris PC.

Contemporary management of congenital malformations of the heart in infants with Ellis - van Creveld syndrome: a report of nine cases. Cardiol Young. 2011 Apr;21(2):145-52. O'Connor MJ, Rider NL, Thomas Collins R, Hanna BD, Holmes Morton D, Strauss KA.

Cognitive and adaptive functioning after liver transplantation for maple syrup urine disease: a case series. Pediatr Transplant. 2011 Feb;15(1):58-64. Shellmer DA, DeVito Dabbs A, Dew MA, Noll RB, Feldman H, Strauss KA, Morton DH, Vockley J, Mazariegos GV.

Hope Growing in Brazil

Eugenia Valadares is a pediatric biochemical geneticist from the Hospital Belo Horizonte in the large Brazilian city of Minas Gerais, home to 22 million people. She is passionately interested in improving newborn screening and medical care for Brazilian children who suffer from genetic disorders. In her country, outcomes for MSUD, GA1 and other conditions are terrible, much as they were for the PA Old Order 25 years ago.

Eugenia was invited to visit by several Brazilian families whose children are cared for at the Clinic. On their own, these parents have formed an impressive consortium of people from their country--parents, physicians, laboratory scientists, government representatives (Ministry of Health), and attorneys--to explore the formation of a non-profit clinic for special children. They have identified Eugenia as a potential Medical Director and she is here to learn as much as she can about our clinic during her 10-day visit. While here, she will spend two days with UPMC transplant team, shadow Dr. Morton and Dr. Strauss in the Clinic and at the hospital, learn laboratory techniques, and begin to formulate an economic plan.

The Clinic will provide guidance, laboratory protocols, and treatment guidelines as they work to develop a clinic of their own.

New Staff

The new voice on the phone is **Erica S. Eisenbise** who joined the Clinic's staff in September as the Office Receptionist. Erica is a recent graduate of Harrisburg Area Community College with an Associates in Science degree and certification as a Medical Assistant. We are all happy to have Erica on board and in the front office to help keep the rest of us organized.

In January the Clinic will welcome **Matthew Sware** as the new Director of Development. Matt, a graduate of Carnegie Mellon University, recently worked at Carnegie Mellon's Development Office in Pittsburgh and is looking forward to returning to his roots near Strasburg to join the Clinic's staff. He will be working to increase support for the Clinic's operating fund through grants and gifts and to increase the Clinic's Research and Education Endowment Fund.

Christine Hendrickson, R.N. and her husband Shawn welcomed their baby, **Everet Louis**, 8 lbs. on October 17th.

At the Clinic - December, 2011
Front row: Dr. Kevin Strauss, Caroline Morton, Dr. Holmes Morton, and Christine Hendrickson with Everet; back row: Dr. Erik Puffenberger, Erica Eisenbise, Adam Heaps, Miriam Echternach, Paul Morton and Donna Robinson. Not pictured, Rebecca Smoker.

We continue our search for a highly motivated, dedicated pediatrician with interest in genetic diseases to join our staff. We thank LGH for their assistance with the search.

Clinic for Special Children
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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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