2015

ANNUAL REPORT

Clinic *for* Special Children

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"Each office visit is like a teaching seminar for us. The doctors explain every detail very thoroughly and help us understand why our children's special needs affect them the way they do."

- LM, *Mother of a patient*

Our Mission

Provide comprehensive

LOCAL MEDICAL CARE,

integrate science and clinical medicine, and

SHARE KNOWLEDGE to improve

the HEALTH OF CHILDREN

who suffer from genetic disorders.

Advancing Science and Medicine,
One Child at a Time.

We are thankful to all of you for another year of service at the Clinic for Special Children promoting healing, community, and medical discovery.

In 2015, the Clinic discovered 32 new known genetic variants that cause disease, and 10 peer reviewed articles in prestigious scientific journals. The work of the Clinic, and the power of genetics, also received national recognition in the November 2015 issue of Scientific American; a testament to the power of science to improve health. Most importantly, however, 2015 marked another year where your faith and support enabled us to leave an indelible mark upon the lives of genetically disadvantaged and underserved children and their families.

The vibrant growth and success of the Clinic for Special Children is a testament to the precious covenant formed between people who need special care and those professionals who are called to serve them. With your continued support, the work of many hands and hearts will safeguard this promise for generations to come.

Kevin A. Strauss, MD
Medical Director

Adam D. Heaps, MS
Administrative Director

Our Vision

We envision the Clinic for Special Children as a MEDICAL HOME

for predominately Amish and Mennonite children who are born with genetic predispositions to disability, chronic disease or untimely death. We continually strive to integrate advanced scientific tools and concepts into clinical practice so that genetically vulnerable children have access to the most timely, affordable and effective healthcare. The Clinic for Special Children represents an innovative and holistic approach to modern medical care that can inform the practice of genomic medicine in other settings. We seek opportunities for education and collaboration that promote the well-being of genetically disadvantaged, underserved individuals throughout the world, and are dedicated to training the young clinicians and scientists who will care for these individuals now and into the future.

GENERAL STATS

1,182 *active patients*

16 *staff members*

Patient Care

Delivering effective and

AFFORDABLE CARE *for* CHILDREN

with genetic conditions.

PATIENT CARE STATS

4,213 *biochemical & genetic tests*

1,575 *patient visits*

Manage 225 *known variants that cause disease*

40 *states &* **17** *countries*

Patient Focus

JULIA WAS DIAGNOSED WITH GLUTARIC ACIDEMIA TYPE I (GA-1) at 5 days of age. GA-1 is a genetic disorder in which the body is unable to process proteins properly. Without proper management, the disorder can cause physical and intellectual disability.

After learning of the diagnosis, Julia's mother was devastated to learn that many children with GA-1 suffer from debilitating effects of the disorder. Unable to find anyone with experience in the disorder locally, Julia's mother began an international search to find a service that could help her daughter. Julia's mother learned of the Clinic for Special Children by searching online. The only problem was Julia's family lived in Hawaii and CSC was located in rural Pennsylvania.

Wanting to ensure the best possible care for Julia, her family moved to Strasburg for what ended up being four years. Fortunately, the Clinic was able to help Julia. "I can't put in words what Dr. Strauss means to us… he helped us in ways you cannot even imagine," says Julia's mother. After Julia grew to an age where a devastating brain injury was much less likely, she and her family were able to move back to Hawaii. Today, Julia is an active, beautiful and vibrant 5 year old who enjoys dancing, swimming, and wants to learn to surf.

"I can't put in words what Dr. Strauss means to us… helped us in ways you cannot even imagine."

Julia's family, originally from Brazil, set up a Facebook page in Portuguese to help connect other families with the services offered by CSC. As a result, patients from Brazil, Portugal, and Spain have been helped by CSC's services.

Education and Community Empowerment

COMMUNITY

The Clinic for Special Children hosts "Family Days" special events where the families of children affected with similar disorders can come together and learn more about the biology of a disorder and the medical treatments in a warm and compassionate environment of fellowship and mutual support. Families from around the country who traveled to Strasburg for the Clinic's Family Day for girls with the DDX3X gene mutation. The families learned about genetic sequencing, the biology behind genetic mutations and more. DDX3X is a genetic mutation that causes a range of intellectual disabilities and in some cases seizures, poor muscle tone or behavioral issues. An estimated 1 to 3% of girls with undiagnosed intellectual disabilities may have this mutation, affecting as many as 1 in 5,000 girls.

EDUCATION

The Clinic, jointly with Franklin & Marshall College, hosted the 3rd Annual conference on Translational Medicine the Plain Populations. The Conference was sponsored by The Children's Hospital of Pittsburgh, Lancaster General Health and Nemours Children's Health System. The conference brought together clinicians, scientists, administrator and other interested individuals to discuss topics related to the Plain populations of North America including: GA-1, MSUD,mental health conditions, therapeutic tissue transplant, cardiac disease and disorders, and early diagnosis and treatment using pulse-ox screening.

EDUCATION & COMMUNITY STATS

10 *family days in 2015*

4 *research interns*

Research

This fall, the Clinic received national recognition by SCIENTIFIC AMERICAN

who featured an article on our own medical director, Kevin Strauss, MD and how genetics can be used to inform medical treatment. Like so many disorders treated at the Clinic, early diagnosis and treatment is essential and may mean the difference between life and death. For one family treated at the Clinic, a SCID diagnosis was made at 4 hours of age and resulted in a life-saving bone marrow transplant at 1 month of age.

READ THE ARTICLE ONLINE AT:

scientificamerican.com/article/clinic-genomics-can-improve-health-care-right-now/

RESEARCH & DEVELOPMENT STATS

32 NEW *variants found that cause disease*

10 *peer-reviewed publications in 2015*

2015 | PUBLISHED PAPERS

1. Strauss KA, Ferreira C, Bottiglieri T, Zhao X, Arning E, Zhang S, Zeisel SH, Escolar ML, Presnick N, Puffenberger EG, Vugrek O, Kovacevic L, Wagner C, Mazariegos GV, Mudd SH, Soltys K. **Liver transplantation for treatment of severe S-adenosylhomocysteine hydrolase deficiency.** Mol Genet Metab. 2015 Jun 18. doi: 10.1016/j.ymgme.2015.06.00.

2. Benkert AR, Young M, Robinson D, Hendrickson C, Lee PA, Strauss KA. Severe salt-losing **3-hydroxysteroid dehydrogenase deficiency: treatment and outcomes of HSD3B2 c.35G>A homozygotes.** J Clin Endocrinol Metab. 2015 Jun 16. doi: jc20152098.

3. Jinks RN, Puffenberger EG, Baple E, Harding B, Crino P, Fogo AB, Wenger O, Xin B, Koehler AE, McGlincy MH, Provencher MM, Smith JD, Tran L, Al Turki S, Chioza BA, Cross H, Harlalka GV, Hurles ME, Maroofian R, Heaps AD, Morton MC, Stempak L, Hildebrandt F, Sadowski CE, Zaritsky J, Campellone K, Morton DH, Wang H, Crosby A, Strauss KA. **Recessive nephrocerebellar syndrome on the Galloway-Mowat syndrome spectrum is caused by homozygous protein-truncating mutations of WDR73.** Brain. 2015 June 12. doi: 10.1093/brain/awv153.

4. Soltys KA, Mazariegos GV, Strauss KA. **Living related transplantation for MSUD–caution, or a new path forward?** Pediatric Transplantation. 2015 May 19. doi: 10.1111/petr.12423.

5. Streeten EA, Ramirez S, Eliades M, Jaimungal S, Chandrasekaran S, Kathleen R, Morton DH, Puffenberger EG, Herskovitz R, Leonard MB. **Fractures on bisphosphonates in osteoporosis pseudoglioma syndrome (OPPG): pQCT shows poor bone density and structure.** Bone. 2015 Apr 16. doi: 10.1016/j.bone.2015.04.007.

6. Yoshikawa M, Go S, Suzuki S, Suzuki A, Katori Y, Morlet T, Gottlieb SM, Fujiwara M, Iwasaki K, Strauss KA, Inokuchi J. **Ganglioside GM3 is essential for the structural integrity and function of cochlear hair cells.** Human Molecular Genetics. 2015 Mar 5. doi: 10.1093/hmg/ddv041.

7. Riley P, Weiner DS, Leighley B, Jonah D, Morton DH, Strauss KA, Bober MB, Dicintio MS. **Cartilage hair hypoplasia: characteristics and orthopaedic manifestations.** 2015 Feb 27. doi: 10.1007/s11832-015-0646-z.

8. Strauss KA, Jinks RN, Puffenberger EG, Venkatesh S, Singh K, Cheng I, Mikita N, Thilagavathi J, Lee J, Sarafianos S, Benkert A, Koehler A, Zhu A, Trovillion V, McGlincy M, Morlet T, Deardorff M, Innes AM, Prasad C, Chudley AE, Lee INW, Suzuki CK. **CODAS Syndrome Is Associated with Mutations of LONP1, Encoding Mitochondrial AAA+ Lon Protease.** American Journal of Human Genetics. 2015 Jan 8. doi: 10.1016/j.ajhg.2014.12.003.

9. Manoli I, Myles JG, Sloan JL, Carrillo-Carrasco N, Morava E, Strauss KA, Morton H, Venditti CP. **A critical reappraisal of dietary practices in methylmalonic acidemia raises concerns about the safety of medical foods. Part 2: cobalamin C deficiency.** Genet Med. 2015 Aug 13. doi: 10.1038/gim.2015.107.

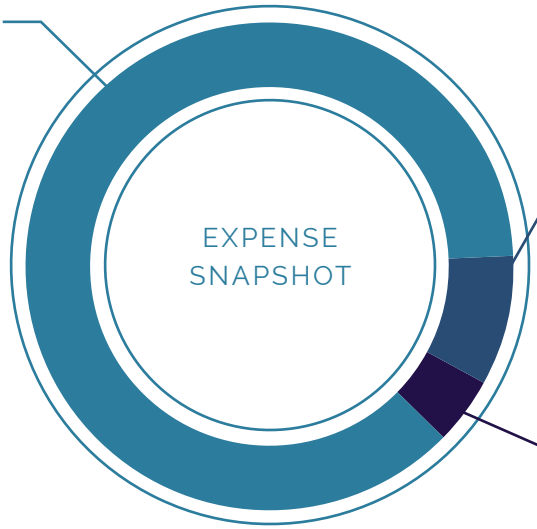
10. Snijders Blok L, Madsen E, Juusola J, Gilissen C, Baralle D, Reijnders MR, Venselaar H, Helsmoortel C, Cho MT, Hoischen A, Vissers LE, Koemans TS, Wissink-Lindhout W, Eichler EE, Romano C, Van Esch H, Stumpel C, Vreeburg M, Smeets E, Oberndorff K, van Bon BW, Shaw M, Gecz J, Haan E, Bienek M, Jensen C, Loeys BL, Van Dijck A, Innes AM, Racher H, Vermeer S, Di Donato N, Rump A, Tatton-Brown K, Parker MJ, Henderson A, Lynch SA, Fryer A, Ross A, Vasudevan P, Kini U, Newbury-Ecob R, Chandler K, Male A; DDD Study, Dijkstra S, Schieving J, Giltay J, van Gassen KL, Schuurs-Hoeijmakers J, Tan PL, Pediaditakis I, Haas SA, Retterer K, Reed P, Monaghan KG, Haverfield E, Natowicz M, Myers A, Kruer MC, Stein Q, Strauss KA, Brigatti KW, Keating K, Burton BK, Kim KH, Charrow J, Norman J, Foster-Barber A, Kline AD, Kimball A, Zackai E, Harr M, Fox J, McLaughlin J, Lindstrom K, Haude KM, van Roozendaal K, Brunner H, Chung WK, Kooy RF, Pfundt R, Kalscheuer V, Mehta SG, Katsanis N, Kleefstra T. **Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling.** Am J Hum Genet. 2015 Jul 28. pii: S0002-9297(15)00280-3. doi: 10.1016/j.ajhg.2015.07.004.

2015 | FINANCES

86%

Program Services

$2,423,597



9%

management

$259,779

5%

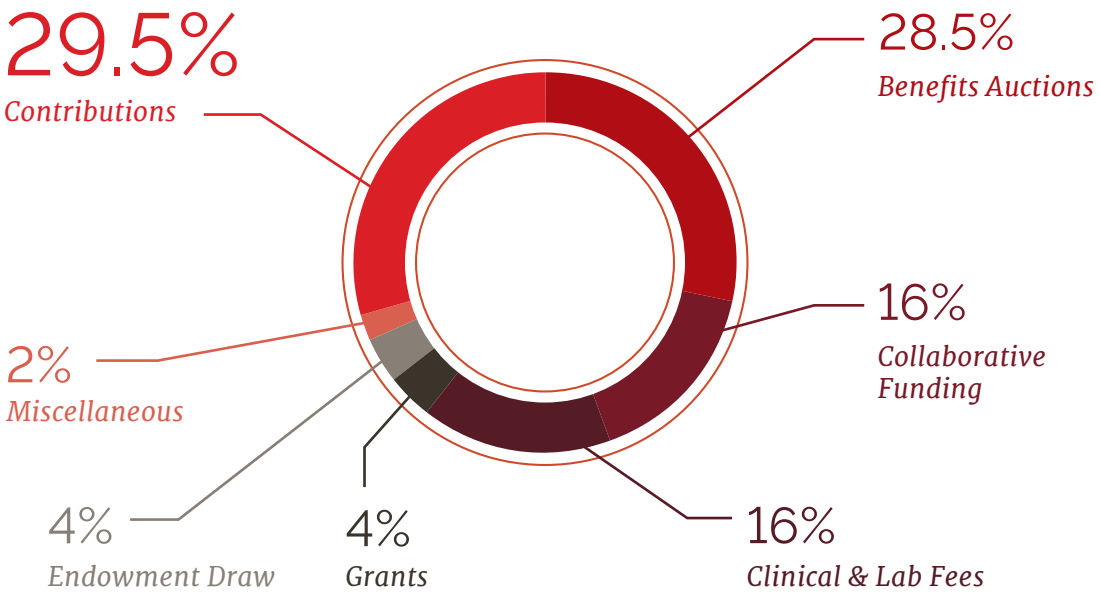
fundraising

$128,791

TOTAL EXPENSES

$ 2,812,167

2015 REVENUE SNAPSHOT



Clinic *for* Special Children

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