Special Page 4 Feature

Act of Faith
Doctors treat infant’s rare genetic Krabbe disease with transplant

Specialty Care Center Opens
Erie gets expanded services

Telemed for Sickle Cell
Connecting kids with treatment

125th Anniversary
Gala celebrates past, future
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The FALL 2015 issue of Pediatric INSIGHTS

Cover story: Early detection can be vital for the successful treatment of rare genetic conditions. Doctors at Children’s Hospital of Pittsburgh of UPMC recently performed an umbilical cord blood transplant on Faith Aleah Galindo to treat her Krabbe disease, making Faith one of the hospital’s youngest cord blood transplant patients at just 23 days old.

In addition:

- For more than two years, a pioneering telemedicine program at Children’s has enabled pediatric patients with sickle cell disease in Erie to access the only disease-modifying therapy currently available.
- Children’s bids a fond farewell to David H. Perlmutter, MD, physician-in-chief and scientific director, who has been instrumental in propelling Children’s Department of Pediatrics into the national spotlight.
- There was a lot to celebrate during the Children's Hospital of Pittsburgh Foundation 125th Anniversary Gala, including an announcement that more than $11.9 million has been raised to date for the anniversary year campaign.

We welcome your feedback, thoughts, and story suggestions. Please share them with one of our physician liaisons, whose contact information you can find on page 7.

Physician Referral Service
412-692-PEDS (7337)

To refer a patient to any of Children’s Hospital of Pittsburgh of UPMC’s clinical services, please call our Physician Referral Service at 412-692-PEDS (7337).

Visit the Referring Physicians section of Children’s website at www.chp.edu/physicians.
New Specialty Care Center Opens in Erie
Convenient Location Offers Expanded Clinical Space for Pediatric Outpatient Services

Children's Hospital of Pittsburgh of UPMC's newest Specialty Care Center, located on the first floor of UPMC Hamot Women’s Hospital, opened its doors on Sept. 8. The new, permanent location offers increased access to Children's Hospital experts for families in the Erie region thanks to increased clinic space.

Pediatric experts from several divisions at Children's travel to Erie on a regular basis to provide outpatient services, including diagnostic and follow-up care to children, adolescents, and young adults.

The Specialty Care Center Erie was originally located at Shriners Hospitals for Children®, and then moved temporarily to CCP-Hamot Pediatrics during the construction of the new location. Children’s continues to offer pediatric orthopaedic services in cooperation with Shriners Hospitals for Children–Erie, 1645 West 8th St., 2nd floor, Erie, PA 16505. For more information, call 814-875-8700.

“Children’s is committed to providing families from Erie with world-class pediatric care that is convenient and close to home,” says Nick Barcellona, vice president and chief financial officer, Pediatric Academic and Clinical Service Line at Children’s Hospital. “We have a large presence in Erie, ranging from our

Express Care Center on Peach Street and three Children’s Community Pediatrics practices in the area, so as the need for pediatric services has increased, we recognized that we needed to move to this new location to accommodate families.”

A ribbon-cutting ceremony for the new Specialty Care Center Erie was held Oct. 15. Children's operates four other Specialty Care Centers in Chippewa, Hermitage, Johnstown, and Wheeling.

For an updated schedule and complete list of providers going to Children’s Specialty Care Center Erie, visit www.chp.edu/erie.
Pioneering Telemedicine Program Brings Care to Sickle Cell Patients

Two daughters with one debilitating disease, and a geographic barrier to the only disease-modifying medication currently available — that's what Mary Brown of Erie had to cope with until two years ago, when Children's Hospital of Pittsburgh of UPMC launched a telemedicine program to connect pediatric patients to sickle cell disease specialists in Pittsburgh.

Sickle cell disease is an incurable, inherited red blood cell disorder. The only proven treatment to reduce painful episodes and costly hospitalizations is drug therapy that requires careful and consistent monitoring by a Pediatric Hematology team.

Debra Cohen, MD, Division of Pediatric Hematology/Oncology; Patricia McLendon, DNP, CRNP; Margaret Holtz, PA-C; Kim Washington, LSW; and genetic counselor Beth Kladny, MS, CGC, travel to Erie to see patients twice annually. Since 2013, monthly telemedicine appointments have allowed a Children's specialist to prescribe the sickle cell drug and remotely monitor the treatment.

"We can't make it to Pittsburgh every month so my girls weren't able to be on the medicine," Mary says. "Now they can be on the medicine because we can see the doctor through telemedicine. The girls have not needed to go to the Emergency Department and have not been admitted to the hospital since we started the telemedicine visits."

The Pediatric Hematology team sees patients at the Children's Specialty Care Center Erie's new location in UPMC Hamot Women's Hospital, 118 East 2nd St., Suite W1150. Children’s will expand the sickle cell telemedicine service to Children's Hospital Specialty Care Center Hermitage in 2016.

The New York Mid-Atlantic Consortium for Genetics and Newborn Screening Services, a federally funded multi-state regional collaborative, has provided funding that will allow Children's to teach up to 80 other health care organizations in Delaware, District of Columbia, Maryland, New Jersey, New York, Pennsylvania, Virginia, and West Virginia how to implement sickle cell telemedicine programs.

To refer a patient to the sickle cell telemedicine service, contact the Division of Pediatric Hematology/Oncology at 412-692-5055.

Pediatric Asthma Center Leads in Care, Research, and Education

The Pediatric Asthma Center at Children's Hospital of Pittsburgh of UPMC has an ambitious vision: To discover and understand the causes of asthma, and to use this knowledge to provide access to state-of-the-art, cutting-edge asthma care to all children in western Pennsylvania and beyond.

In the Pediatric Asthma Center, experts on asthma, lung function, breathing sensitivities, and the immune system team up to provide the highest level of care for patients. Earlier diagnosis and more effective management of young children with asthma decrease the number of cases of undiagnosed asthma and reduce the number of emergency room visits for children with asthma. For pediatric patients with severe asthma, the Difficult-to-Treat Asthma Clinic provides a dedicated clinic space where children are seen by both a pediatric pulmonologist and a pediatric allergist. Of 31 children seen since the clinic’s establishment in 2012, 19 (61 percent) reported significantly fewer asthma attacks by June 2014.

In the research arena, Pediatric Asthma Center specialists conduct both clinical research studies as well as basic research to investigate the impact of environmental and lifestyle factors on children. Current human studies of asthma funded by the National Institutes of Health include Epigenetic Variation and Childhood Asthma in Puerto Ricans; Genes, Allergens, and Asthma in Puerto Ricans; and Vitamin D to Prevent Severe Asthma Attacks.

Community outreach and education is another important tenet. Asthma specialists team up with the Ronald McDonald Care Mobile to visit three schools in the Woodland Hills School District, providing education, clinic visits, and pulmonary function testing for children with asthma who cannot reach a Children’s Hospital location easily. The Care Mobile visits each school twice per school year. Physicians from the Pediatric Asthma Center also are available to give talks in the community.

Asthma testing and appointments with an asthma specialist are available at all Children's Hospital locations. To contact a physician, call Melisa Kennedy, administrative coordinator, at 412-692-8736.
Pediatric Pathology Continues to Advance Treatment for Children

Pathology — the study of the origin, nature, and course of disease — forms the backbone of modern medicine. The Division of Pediatric Pathology at Children's Hospital of Pittsburgh of UPMC is one of the nation’s oldest and most prestigious pediatric pathology departments, and today its members continue to lead in the field.

“For many diseases, physicians can’t begin to treat a child without receiving a formal diagnosis from Pathology,” says Miguel Reyes-Múgica, MD, chief of Pathology and head of Laboratories at Children’s, and editor-in-chief of Pediatric and Developmental Pathology, the journal of the Society for Pediatric Pathology and the Paediatric Pathology Society. “Pathologists learn to see what is happening in a child's body and interpret it. We’re physicians, not merely technicians.”

State-of-the-art clinical and anatomic pathology laboratories at Children’s Hospital enable pathologists to provide a full range of consultative and diagnostic services including automated testing technologies, routine histopathology, molecular diagnostics, cytology, immunohistochemistry, in situ hybridization and ultrastructural pathology, microbiology, and virology — all of which are vital in guiding the diagnosis and therapy of Children’s patients.

Each year, Pediatric Pathology analyzes more than 11,000 surgical specimens and 2,000 cytological samples, and performs approximately 1 million lab tests and 40 autopsies.

Maud Menten — best known for the Michaelis-Menten equation for calculating an enzymatic reaction — led Pathology at Children’s, and taught at the University of Pittsburgh School of Medicine from 1923 to 1950. To this day, the highest honor in the Department of Pathology at the University is the Maud L. Menten Professor of Pathology.

The Frank E. Sherman and Cora C. Lenox Heart Museum houses more than 2,700 congenitally malformed hearts studied by researchers from around the world. Frank Sherman, MD, started the museum in 1954. Cora Lenox, MD, came to Children’s in 1959 and re-catalogued all of the heart specimens.

Doctors continue to make advances in the following areas:

> Amy Davis, MD – vascular anomalies and hemangiomas
> John Ozolek, MD – head and neck pathology, and advanced morphometric techniques for diagnosis
> Jennifer Picarsic, MD – heart and lung pathology, and histiocytic disorders
> Sarangarajan Ranganathan, MD – pediatric neoplasia, particularly liver tumors
> Miguel Reyes-Múgica, MD – pediatric neoplasia, neural crest diseases, disorders of sex development, pediatric testicular pathology, and congenital melanocytic lesions

To consult with a member of the Pediatric Pathology Division, call 412-692-5650.
Both children were born with Krabbe disease, a devastating genetic disorder. Although Zarely’s diagnosis came too late to save her life, because of her, Faith’s parents knew what to do following a prenatal diagnosis to give Faith every chance to lead a full life.

At 23 days old, Faith became one of the youngest patients to receive an umbilical cord blood transplant at Children’s Hospital of Pittsburgh of UPMC, where a unique, dedicated program in leukodystrophies led by Maria Escolar, MD, MS, draws patients from across the United States and around the world.

What is Krabbe disease?

Krabbe disease (pronounced “Crab-A”), also called globoid cell leukodystrophy, is caused by a deficiency in the enzyme galactocerebrosidase, or GALC, that impairs the growth of myelin, a substance that forms the myelin sheath. That sheath covers and protects nerve cells like a layer of insulation; without it, nerve cells degenerate. In early-onset infantile Krabbe disease — the most common form — symptoms such as irritability, loss of muscle tone, and seizures usually appear by age 6 months. The disease, which affects about one in 100,000 people in the United States, is inherited in an autosomal recessive pattern (both parents carry a copy of the faulty
gene, although they typically have no signs or symptoms of the condition).

For the first few months following her birth in 2005, Zarely appeared to be a normal, healthy baby, recalls her mother, Yesenia Galindo-Lopez. She was a bit fussy, which was attributed to colic, but she fed normally and was meeting her developmental milestones. At 5 months, however, she began to regress. She could no longer sit up or hold up her head. She cried inconsolably. Her body began to stiffen. The family’s pediatrician referred her to a neurologist, but before the date of that appointment arrived, Zarely was hospitalized with a seizure.

Neither Yesenia nor her husband, Raul, had heard of the disease their daughter was diagnosed with — Krabbe. Zarely died in 2007, two years after her diagnosis. The following year, at a meeting of a support and advocacy organization for families of children with Krabbe, Yesenia met Dr. Escolar.

**Success with transplantation**

An internationally renowned expert on leukodystrophies (a group of rare genetic diseases involving impaired myelin), Dr. Escolar has led the Program for the Study of Neurodevelopment in Rare Disorders at Children’s Hospital since 2011. She authored the first scientific paper reporting the successful transplantation of umbilical cord blood in babies with infantile Krabbe disease, published in the *New England Journal of Medicine* in 2005. In this study, all asymptomatic infants who received transplants survived and developed age-appropriate cognitive function and language skills, although a few experienced some delays in language and motor function. By contrast, infants who received transplants after the onset of Krabbe symptoms were less likely to survive and had minimal neurologic improvement.

“The brain myelinates very quickly during the first year of life,” Dr. Escolar explains. “We know that Krabbe disease can begin to progress in utero. Transplantation halts the disease’s progression but cannot reverse damage that has already occurred. So it’s important to perform the transplant as early as possible — every day makes a difference.”

**Coming to Pittsburgh**

Yesenia Galindo-Lopez gave birth to a healthy daughter, Chloe, in 2009. Prenatal testing had shown that Chloe was a carrier of the faulty gene for Krabbe but did not herself have the condition. When Yesenia became pregnant again in 2014, prenatal testing revealed that this baby, also a girl, did have Krabbe.

The Galindos decided to name the baby Faith Aleah. “Faith is believing in what is unknown,” says Yesenia. Aleah comes from a Hebrew word meaning...
“to ascend.” Then Yesenia called Dr. Escolar.

A cord blood transplant involves several weeks of hospitalization followed by several months of close follow-up care. It was a daunting decision for the Galindos to move the family from their home near Los Angeles to Pittsburgh for Faith’s treatment. In addition to Chloee, now 5, they have an older daughter, Isabel, 13.

“We felt that Faith would have the best chance for a good outcome here,” says Yesenia. “But it was also important to us to keep the family together.” Isabel and Chloee are staying at Ronald McDonald House with Yesenia’s mother, Teresa Lopez, while Yesenia stays in the hospital, where she is nursing Faith. Following the transplant, dad Raul returned to California for his job, but flies across the country to visit Pittsburgh as often as he can.

Facing the future
Just 13 days following Faith’s transplant, blood tests showed that she had fully engrafted — 100 percent of the blood cells in her body could be identified as originating from the donated cord blood. During the transplant process she grew two inches, gained about 1¾ pounds, and continued to nurse or receive breast milk in a bottle.

“She has avoided most of the dangerous complications associated with the immediate post-transplant period,” says Paul Szabolcs, MD, chief of Children’s Division of Blood and Marrow Transplantation and Cellular Therapies. He expects that Faith will be able to go home to California with her family in early December and well enough to discontinue immunosuppressant medication by May, nine months after her transplant. She and her mother will return to Pittsburgh two to four times a year for follow-up visits with Dr. Escolar.

Whatever the future holds for Faith, Yesenia says she is comforted by knowing that her daughter is being cared for by doctors with a wealth of experience treating patients with Krabbe disease.

For an appointment, consultation, or referral for a patient with Krabbe disease or another form of leukodystrophy, contact the Program for the Study of Neurodevelopment in Rare Disorders at Children’s Hospital at 412-692-9955. Visit us online at chp.edu/nrdrd.
Basketball is a game of fast-moving variables. Winning teams are distinguished by their informed strategy, trusting partnerships, and ability to quickly adapt to shifting forces.

Here at Children’s Hospital of Pittsburgh of UPMC, David H. Perlmutter, MD, physician-in-chief and scientific director, has long been an unabashed fan and observer of the game. Like the greatest of coaches, he’s a strategic thinker who understands change, takes calculated risks, and nurtures the unique talents of those with whom he works.

As Children’s Hospital celebrates its 125th anniversary and we reflect on the legacies of the many individuals whose contributions helped transform our hospital — and shape our winning team — it’s clear Dr. Perlmutter’s name will long be invoked among the “best of the best.”

Since his arrival here in 2001, Dr. Perlmutter has been instrumental in propelling the University of Pittsburgh School of Medicine’s Department of Pediatrics into the national spotlight. A physician-scientist who is one of the world’s foremost experts on the rare disease alpha-1 antitrypsin (AT) deficiency, he has led a five-fold increase in National Institutes of Health (NIH) funding from $8 million to nearly $40 million today. For the last six consecutive years, U.S. News & World Report has ranked Children’s among the top 10 pediatric hospitals in the country.

In a 2008 interview, Dr. Perlmutter said his goal for Children’s Hospital was “to provide outstanding clinical service and to excel in education and research.” The ability to balance that trinity effectively is one that eludes many administrators, but Dr. Perlmutter was relentless in his efforts to recognize the critical importance of each.

Two years ago, UPMC invested $30 million to support research excellence at Children’s Hospital, including the creation of the $10 million David H. Perlmutter Research Endowment that enables us to continue to recruit and retain national leaders in pediatric research.

Our patients and their families are the greatest beneficiaries of Dr. Perlmutter’s leadership. Our medical, academic, and regional communities also have reaped many rewards from Children’s continued growth and innovation.

At Children’s Hospital of Pittsburgh of UPMC, research and clinical care go hand in hand. We believe the most important discoveries in the laboratory are the result of exploring ideas born from our experience in treating the children who come to us for care.”

— David H. Perlmutter, MD

Dr. Perlmutter will leave Children’s Hospital on Dec. 1, 2015, to assume the role of executive vice chancellor for medical affairs and dean of Washington University School of Medicine in St. Louis, where he earned his medical degree. It’s a remarkable opportunity for him to return to one of the country’s most highly ranked medical schools and research programs — a university that he and his wife, Barbara Cohlan, MD, know well and love.

Although we say good-bye with heavy hearts, we wish David and Barbara well. And, we offer our most sincere thanks for leaving us with a legacy of excellence.

Andy Urbach, MD, is associate chief medical officer at Children’s Hospital. He welcomes your comments and questions. Please send an email to mdrelations@chp.edu.
Laurels

These Children’s Hospital staff members recently received the following recognition in their fields.

Diego Chaves-Gnecco, MD, PHD, (second from right) received the NFL Hispanic Heritage Award on behalf of the Steelers on Sept. 20. Dr. Chaves-Gnecco is director and founder of Salud Para Niños (Health for Children) at Children’s Hospital of Pittsburgh of UPMC as well as a developmental-behavioral pediatrician. At Salud Para Niños, culturally and linguistically competent primary care for children and families is complemented with activities that focus on prevention and community empowerment. Dr. Chaves-Gnecco also is assistant professor of Pediatrics at the University of Pittsburgh School of Medicine.

Alene D’Alesio, DMD, of the Division of Pediatric Dentistry at Children’s Hospital of Pittsburgh of UPMC, was named one of America’s 40 top young dentists under age 40 in the fall 2015 issue of the magazine, Incisal Edge. She is the inventor of the Lily Method, a five-step pacifier system to wean toddlers in as few as five days. The product won a gold medal at the 2015 Edison Awards.

Maria Escolar, MD, MS, director of the Study of Neurodevelopment in Rare Disorders Program, was named a 2015 RARE Champion of Hope for Medical Care and Treatment. The award is presented by Global Genes, an international rare disease patient advocacy organization that annually honors individuals and organizations for extraordinary efforts in advocacy, science, awareness, medical care and treatment, industry, documentary, and brave feats of courage.

Pittsburgh-area Hyundai dealers and Hyundai Hope On Wheels presented J. Anthony Graves, MD, PhD, a cancer researcher at Children’s Hospital of Pittsburgh of UPMC, with the 2015 Hyundai Scholar Senior Researcher Award. The $250,000 grant will support Dr. Graves’ research of neuroblastoma, a solid tumor that usually develops in the nerve tissue of the adrenal glands, located on the top of each kidney. Dr. Graves is assistant professor of Pediatrics at the University of Pittsburgh School of Medicine.

Amy Houtrow, MD, PhD, MPH, co-authored a new report from the National Academies of Sciences, Engineering and Medicine that found many low-income children with mental disorders who are eligible for federal benefits may not be receiving them. The report, “Mental Disorders and Disabilities Among Low-Income Children,” also noted that the number of children who do receive assistance has been rising in accordance with overall mental health trends and rising poverty rates. Dr. Houtrow, chief of the Division of Pediatric Rehabilitation Medicine at Children’s Hospital of Pittsburgh of UPMC, served on the committee that authored the report. She is associate professor of physical medicine and rehabilitation and pediatrics at the University of Pittsburgh School of Medicine.

The book Handbook of Pediatric Cardiovascular Drugs, Second Edition, edited by Ricardo Munoz, MD, was selected by the British Medical Association as among its six most recommended books in cardiology. Dr. Munoz is chief of Cardiac Intensive Care, director of the Cardiac Recovery Program, and medical director for Global Business and Telemedicine at Children’s Hospital of Pittsburgh of UPMC. He also is professor of Critical Care Medicine, Pediatrics, and Surgery at the University of Pittsburgh School of Medicine.

Edward Prochownik, MD, PhD, director of oncology research at Children’s Hospital of Pittsburgh of UPMC, and the Paul C. Gaffney Professor of Pediatrics at the University of Pittsburgh School of Medicine, was awarded a research grant of $100,000 from the St. Baldrick’s Foundation, a volunteer-driven charity dedicated to raising money for childhood cancer research. The award to Dr. Prochownik is one of 70 grants totaling more than $21.1 million nationally and internationally awarded by St. Baldrick’s in support of pediatric oncology research. These grants provide resources to institutions to conduct more research and enroll more children in ongoing clinical trials. Dr. Prochownik and his team will explore the implications of new observations of cancer cell growth.

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Children’s Hospital of Pittsburgh Foundation held a black-tie gala on Friday, Oct. 2, to celebrate the 125th anniversary of Children’s Hospital of Pittsburgh of UPMC, and announced that $11.9 million has been raised to date during the anniversary year campaign.

The 125th Anniversary Gala was held at the David L. Lawrence Convention Center with more than 900 supporters, including celebrities, athletes, and business and government leaders. The event celebrated all of the community support that has enabled Children’s Hospital to maintain its mission since 1890.

“This celebration recognizes 125 years of caring by our community and the hundreds of thousands of individuals who have given their time and support to ensure that every child in this region has access to the care they need and deserve,” says Greg Barrett, president, Children’s Hospital of Pittsburgh Foundation.

The honorary chair for the evening was actor and Pittsburgh native, Joe Manganiello. Emcee for the gala was KDKA-TV anchor Ken Rice. Other celebrity guests included former Pittsburgh Steelers Brett Keisel and Troy Polamalu; Mario Lemieux, founder, Mario Lemieux Foundation; and Stephanie McMahon, chief brand officer, WWE.

“The dedication, commitment and generosity of the people of Pittsburgh and the surrounding communities has been and will continue to be the driving force behind Children’s,” says Christopher Gessner, the president of Children’s Hospital of Pittsburgh of UPMC. “This tremendous community support enables our expert physicians, nurses, and other staff members to provide the best pediatric health care in the world every single day.”

The 125th Anniversary Gala honored Beacon of Hope recipients Eat’n Park Hospitality Group and Howard Hanna Real Estate Services, which collectively have raised nearly $20 million. Legacy of Hope recipient John G. Rangos Sr. is a renowned business leader and member of the Children’s Hospital of Pittsburgh Foundation Board of Trustees. He has donated more than $9.5 million to Children’s, including a significant gift in 1989 that helped to found the first Children’s Hospital research building.

Denise Pampena and Ramsey Lyons, who serve as trustees, co-chaired the event.

Children’s Hospital began its 125th celebration with a kickoff celebration on June 4 that launched the “Give Kids a Chance to Be Kids” campaign — celebrating 125 years of caring and the important role of community support for the clinical and research advances at Children’s. The anniversary year will culminate on June 4, 2016, with Walk for Children’s, a new grassroots community fundraising event.

PICTURED (top to bottom): Joe Manganiello, honorary chair; performance by Cirque Mechanics; Sam Cain, Children’s Hospital patient and honored guest; Greg Barrett, Foundation president, and Christopher Gessner, Children’s Hospital president.
GIVE
KIDS AN CHANCE TO BE KIDS

visit givetochildrens.org/125