When an illness has no name

A HOCKEY INJURY LEADS TO A STARTLING DISCOVERY

IF YOU GIVE A KID A KALE CHIP...
Letter from the CHAIR

Dear Friend of Duke Children’s,

Teenager Colby Heath suffered a hockey injury that left him with no feeling in his legs. He recovered quickly and later walked out of the emergency department. But this was only the beginning of his journey: Months later, his family was blindsided by what they learned. Read his amazing story starting on page 10.

Young Issac Locklear graces our cover. Issac’s perplexing symptoms presented a mystery to his doctors. Thanks to our brilliant physicians and Duke Children’s link to the Undiagnosed Diseases Network, Issac’s parents now know the cause (and surprising name) of his condition. On page 4, read more about this family and another who got answers, thanks to the work of these physicians.

And don’t miss the story on page 8 about a fantastic obesity-prevention program for kids. In Durham, the program is called Bull City Fit, and Duke Children’s will replicate its successful model in sites across North Carolina, thanks to a grant from The Duke Endowment.

Enjoy the spring and this issue of Stories!

Because nothing matters more,

ANN M. REED, MD
Chair, Department of Pediatrics
Physician-in-chief, Duke Children’s

HOW YOU CAN HELP

You can support lifesaving research and patient care at Duke Children’s. Make a gift any of these ways:

1. Use the envelope bound into this newsletter
2. Use the secure online form at gifts.duke.edu/dch
3. Call us at 919-385-3138
Duke One of Five Leading Sites in Nation for Children’s Surgery

Duke Children’s is one of only five children’s surgical centers in the nation certified Level 1 by the American College of Surgeons. This certification recognizes the capability to provide the most complex care and services for patients and families. To achieve Level 1 certification centers must offer multiple medical and surgical specialties, including those that are uncommon or require substantial multidisciplinary coordination.

“This is a significant milestone not only for Duke Children’s but also for Duke Health,” said Jeffrey R. Marcus, MD, chief of surgery at Duke Children’s. “This designation underscores our ongoing commitment to providing the finest possible care to all of our patients. It is a source of great pride for our entire team, but the true winners are the children who will benefit from the changes that were required to reach this goal.”

OUTCOMES AT DUKE CHILDREN’S NICU OUTPERFORM NATIONAL AVERAGES

The neonatal intensive care unit (NICU) at Duke Children’s is outperforming comparable international centers in rates of mortality, chronic lung disease among survivors, and mortality, survival, or diagnosis with morbidity during NICU stays, according to data collected by the Vermont Oxford Network.

“For high-risk neonates, time in the NICU is critical for surviving without morbidity, as is the continuity in care received by the mother and neonate,” says neonatologist Ronald Goldberg, MD. “These numbers show that premature neonates fare better at Duke.”

Duke Children’s neonatal program delivers robust assessment, support, and care, providing follow-up care for the first year of life or beyond. The program has added palliative care capabilities and is developing cardiology and neurology units. With six beds already designated for neonates with neurologic issues, Duke’s NICU is one of only a handful of “neuro-NICUs” in the United States.
POTENTIAL TREATMENT FOR PRADER-WILLI SYNDROME

Duke researchers have identified a drug-like molecule that, in animal experiments, appears to be an effective treatment for Prader-Willi syndrome. Prader-Willi syndrome is a genetic disorder that is characterized by poor feeding, growth, and weak muscles in infancy, followed by excessive eating, obesity, and behavioral problems in childhood. It occurs in about one of every 15,000 births and has no cure. If the findings by the Duke-led team uphold in human studies, the drug could become the first treatment option for Prader-Willi syndrome. Yong-hui Jiang, MD, PhD, associate professor in Duke’s departments of pediatrics and neurobiology, is senior author of the study.

Faculty Honors

Sallie Permar, MD, PhD, has been named the new director of the Duke Pediatric Research Scholars Program for Physician-Scientist Development. Rasheed Gbadegesin, MD, has been named the associate program director. In these new roles, Permar and Gbadegesin will work to recruit and retain top trainees with records of success in clinical and basic research, and will support their dual interests during and after residency and fellowship training.

In addition, Permar has accepted the position of director of the Duke Children’s Discovery Institute. In this role, she will lead the effort to discover and develop a research program focused on the early influences of health and disease.

DUKE PEDIATRICS LEADS IN NIH FUNDING

The Duke Department of Pediatrics has been ranked third nationally on the list of National Institutes of Health funding for pediatric clinical science departments. Pediatric investigators in the department received a total of $38,935,527 in grant funding for 2016. The Department of Pediatrics was ranked fifteenth in total funding in 2015. The rise in rankings reflects the groundbreaking work of the faculty, staff, and researchers in the department.
2016 Duke Children’s Gala Grant Recipients

The 2016 Duke Children’s Gala was held October 2016. Duke Children’s physicians received grants ranging from $25,000 to $30,000 resulting from funds raised during the Gala. Grants were awarded to physician projects focusing on care for the complex patient.

The 2016 recipients are:

Richard Chung, MD – Development of an Integrated Pediatric Complex Care Continuum

Karen Jooste, MD – PedsTalk: Improving Physicians’ Communication Skills for Difficult Conversations and for Delivering Serious News to Pediatric Patients and their Families

Gary Maslow, MD – Parent Navigators to Provide Support for Children and Youth with Neurodevelopment Disorders

Victoria Parente, MD – The Duke Children’s “CARE” (Caring for the Region’s Exceptional Children) Network: identifying community needs for children with medical complexity and high resource utilization

Carolyn Pizoli, MD, PhD – Duke Children’s Cardiac NeuroDevelopmental Outcomes Program (CaNDO)

Heather Van Mater, MD – Rheumatology Patient Navigator

YOUR SUPPORT IS MAKING A DIFFERENCE!

SPRING 2017 3
ON THE TRAIL OF UNKNOWN DISEASES
DUKE DOCTORS GIVE FAMILIES LONG-SOUGHT ANSWERS

BY DAVE HART
IT WASN’T UNTIL QUINN WAS ABOUT FIVE MONTHS OLD THAT HIS PARENTS, LIZ ARONIN AND JAMIE MILLS, STARTED TO SUSPECT THAT SOMETHING WASN’T QUITE RIGHT. At an age when most children begin to sit up, to grasp objects, to babble, to track visual movement and respond to voices, Quinn didn’t do any of those things.

“His doctor initially said to wait, because kids develop at different rates,” says Jamie, a grants administrator at Duke. “But at about seven months, she said, ‘OK, I’m concerned now too.’”

Months of appointments, tests, and meetings with specialists soon followed. Meanwhile, Quinn needed constant care. He had seizures. He cried for entire days. Liz and Jamie traded shifts, but there was never enough sleep, never a respite from the stress of knowing their child was seriously ill, but not knowing why.

“We were just surviving,” says Liz, a Duke nurse. “Quinn was so miserable I couldn’t even take him out of the house. You’re sleep-deprived, housebound, and you don’t know what’s wrong with your child. We were really struggling to take care of Quinn and ourselves.”

At the end of all those tests, Liz and Jamie still didn’t know what Quinn had. And they didn’t know whether they could ever have more children without risk.

“Then our genetics counselor said, ‘Have you heard of the Undiagnosed Diseases Network at Duke?’” Jamie says.

AT A GLANCE

- Two families searched for answers to their children’s mysterious illnesses.
- Both were diagnosed with previously unnamed diseases.
- The Undiagnosed Diseases Network links Duke with six other academic medical institutions around the nation to share information about rare, unidentified diseases.
- When diseases are identified, parents have peace and doctors have targets for therapy.
Cracking a Mystery

The Undiagnosed Diseases Network (UDN) is a National Institutes of Health initiative that links Duke and six other academic medical institutions around the nation to share information about rare, unidentified diseases.

The Duke UDN team, led by Vandana Shashi, MD, a professor of pediatrics in the Division of Medical Genetics, set to work to try to solve Quinn’s mystery.

Shashi re-analyzed Quinn’s genetic and clinical information. Drawing on the UDN’s database, she zeroed in on a variant in a gene called NACC1.

With the assistance of Duke UDN genetic counselor Kelly Schoch, similar patients were soon found. “We found six other patients around the world with the same genetic variant and similar clinical presentations,” says Shashi.

NACC1 had never before been known to cause illness, but the evidence was unmistakable. Shashi could give Liz and Jamie and the other six families a diagnosis at last: their children had a previously unknown disease caused by an NACC1 mutation.

A Degree of Peace

Just knowing what was wrong with Quinn made a big difference. So did being able to communicate with the other families.

“It was so satisfying to talk with other people who are going through the same thing,” says Jamie. “And some of the kids are much older, so that gives us some idea of what to expect.”

Maybe the biggest difference for Liz and Jamie was that, since they don’t carry the NACC1 mutation, they felt secure in growing their family. Their second child is due this June.
“Just getting an answer provides a degree of peace that you can’t imagine if you’ve not been there,” Shashi says. “But getting an answer is also the first step toward therapeutics. And that’s what we’re most interested in.”

A Daily Challenge
Shashi took on Quinn’s case not long after tackling another mysterious disease with another Duke UDN investigator, Loren Del Mar Pena, MD, PhD. This one involved a family from Sanford, North Carolina.

Teresa Locklear’s son Issac had a mystifying range of symptoms, including mild developmental delays, cardiac and GI issues, poor motor skills, large size, and a distinctive birthmark on his forehead. Life was a daily challenge.

“People say, ‘How do you do it?’” says Teresa. “I tell them you take it one day at a time. And when you feel like you can’t make it through a whole day, you take it an hour at a time.”

Like Quinn, Issac had seen numerous specialists without a diagnosis before finding the UDN at Duke. Every line of evidence Shashi and Pena unearthed pointed to a mutation in a gene called ASXL2. Through the UDN, they found five more children throughout the world with the same condition.

In 2016, when Issac was 8, Schoch called Locklear to tell her the doctors’ diagnosis: a newly discovered genetic disease, now known as Shashi-Pena Syndrome.

Doors of Discovery
For Shashi and Pena, the UDN has opened new doors of discovery, and new targets for therapy. “Genetics has moved from purely diagnostic into a therapeutic space,” says Pena. “In the end, that’s what we’re after. Whether we’re in the clinic or on the research side, we’re always looking for ways to help our patients.”

How to make a gift to Duke Children’s
You can help Duke Children’s provide answers for more families. To make a donation to Duke Children’s go to gifts.duke.edu/dch
THE DUKE ENDOWMENT FUNDS REPLICATIONS OF A SUCCESSFUL OBESITY PREVENTION PROGRAM FOR CHILDREN

TOWARD A HEALTHIER FUTURE FOR OUR KIDS

BY ASHLEY STRAHM
LIKE MANY TEENS, KAMERON HORTON OF DURHAM LOVES TO PLAY BASKETBALL. But for Kameron, basketball has been more than just a game; it has helped him through serious life challenges.

Kameron lost his father a year and a half ago. At the same time, Kameron, at just 14, was facing concerning health issues, including high glucose, blood pressure, and BMI.

Kameron and his mother, Teresia Parker, were referred to Duke Children's Healthy Lifestyles program by their primary care doctor. The program’s partnership with Durham Parks and Recreation created Bull City Fit, which aims to reduce obesity and improve health among vulnerable populations through diet and exercise.

Basketball became not only Kameron’s go-to workout, but a way to heal.

Through the program, Kameron bonded with older male mentors. “Kam needed to be around other males – good, positive role models,” says his mother. “Being out there and being able to interact and play basketball with those guys was important.” An opportunity to attend a Duke women’s basketball game and shoot during halftime was huge for her son, now a freshman at Southern High School.

The impact on his physical health has been positive as well. “His weight has come down and his BMI is great,” Parker says.

Gabriela Maradiaga Panayotti, MD, is the new director of the Healthy Lifestyles program. Her role presents a unique opportunity to effect change in Durham’s multicultural landscape. Born in Honduras and raised in Central America, she is passionate about working with Hispanic communities. “It allows me to do the medical part I like, but work in the context of cultural familiarity that I really enjoy.”

Maradiaga hopes to circumnavigate challenges to care: social and language barriers, access to safe places to exercise, and easy access to healthy foods.

In late 2016, The Duke Endowment awarded a $749,000 grant to Duke Health to support the replication of Bull City Fit among vulnerable populations outside the Durham area.

“It was great to feel the validation of The Duke Endowment grant,” says Maradiaga. “It means people are really listening and reinvigorating us to keep branching out to serve children in surrounding communities. Now, we have the resources to make it happen.”

The foundation of the program was built over the course of 10 years, in large part by Duke Children’s Sarah Armstrong, MD.

Parker says the program provided positive health outcomes for both Kameron and herself. “Both Kameron and I can exercise and be active together.”

WANT TO KNOW MORE?
Ask your child’s primary care doctor if your family could benefit from Duke Children’s Healthy Lifestyles program. You can support this program with a gift at gifts.duke.edu/dch.
SKATING ON THIN ICE
A TEEN’S LIFE-THREATENING CONDITION IS DISCOVERED ONLY AFTER A FALL DURING A HOCKEY GAME

BY BURGETTA WHEELER
That Colby Heath slept a lot hardly concerned his mother. He was a growing teenage boy after all. And though Colby’s occasional nosebleeds were a nuisance, they weren’t in any way worrisome.

Yet, as Colby’s family now knows, those were two clues that something was horribly wrong.

“He went from being a seemingly healthy kid who plays hockey to all of a sudden a kid who needs a liver transplant in a very short span of time,” says Megan Butler, MD, a hepatologist who has treated Colby at Duke Children’s.

In fact, it was playing hockey that likely saved Colby’s life. During a game early in 2015, a body check sent Colby to the ice, where he alarmingly remained. He couldn’t feel his legs, he said. Taken to an emergency department, he eventually regained sensation and walked out of the hospital that night.

At his hockey evaluations that fall, however, he reported that he was still having occasional numbness in his legs. His parents, Naomi and Brian Askew of Apex, North Carolina, are both emergency department nurses, and they immediately had him evaluated at a Wake County hospital.

Tests showed that Colby had a rare congenital liver abnormality. His portal vein was missing, and his body had responded by creating new pathways for blood that resulted in a dangerous increase in pressure in his blood vessels. That pressure caused his spleen to enlarge and the veins in his esophagus and elsewhere to swell in ways that were potentially life-threatening.

“Colby was at risk of a ruptured spleen and possible death with any abdominal injuries,” Butler says. “Playing hockey was obviously a risk factor.”

**AT A GLANCE**

- Colby’s rare liver abnormality was discovered only after a hockey injury sent him to the hospital.
- Because of the seriousness of the condition, he was referred to Duke Children’s, where doctors determined only a transplant would save his life.
- After months on the transplant list, the family finally got the call that a liver was available.
- Last Christmas, Colby bought toys and gave them to patients at Duke Children’s. One day soon, he hopes to return to the ice as a player.
Delay of game

Because of the seriousness of the condition, Colby was referred to Duke Children’s, where the team determined that only a new liver would save Colby’s life. His scarred liver was failing to remove toxins from his blood, causing ammonia to build up periodically in his brain, a condition called hepatic encephalopathy. Fogginess, confusion, and sleepiness often result.

In February 2016, Colby was placed on the United Network for Organ Sharing list, added to the names of 86 other North Carolina children under the age of 18 waiting for an organ donation. Duke performs 16 to 18 pediatric liver transplants each year.

Much to Colby’s unhappiness, the enlarged spleen meant that he couldn’t play hockey while he waited. At the rink, he started working as a scorekeeper instead. “He has a huge, strong work ethic like no other,” says Naomi Askew, mother also to Kristyn Heath, 20, and Carter Askew, 3.
The call

The day the family got the call that a liver might be available is hard for Colby to remember. About 10 days before, he had to make a trip to the ER, where his ammonia level was found to be 161 micrograms per deciliter. The high end of the normal range is about 50.

His mother, however, remembers May 22, 2016, vividly. Colby was asleep at the family’s home in Apex. Naomi was at a hair appointment 20 miles away and didn’t feel her phone vibrate.

When she returned the call to David Cousino, the pediatric transplant coordinator whom the family uniformly adores, he joked about her missing the call. “Then he said, ‘Is today a good day to get a liver?’” Naomi recalls. “I just stopped. I’m screaming in the middle of the salon, saying, ‘Yes! Yes! Yes!’”

Colby’s surgery would not be a straightforward transplant. Because of the teenager’s extremely rare blood vessel issues, even more exacting precautions and skills would be needed. Twelve long hours later, surgeon Stuart Johnston Knechtle, MD, let the family know that they were finished and that Colby was doing well.

Taking stock

“I can’t thank Duke enough,” Naomi says. “We love them.” Her gratefulness also extends to the family who reached through their grief to donate a loved one’s organs. “How do you thank somebody for a gift you can’t put a price tag on?” Naomi asks.

Since the transplant, Colby, now 15, has donated the hair he started growing at the beginning of his odyssey to the Children With Hair Loss nonprofit. At Christmas, he bought toys with the money earned at the rink and gave them to patients at Duke Children’s. One day soon, he hopes to return to the ice as a player.

Having a new liver should not keep Colby from his sport, Butler says, and she is eager to see how he progresses. “That’s one of the great things is we get to see patients in the clinic years down the line, hear how they’re doing in school, about boyfriends and girlfriends, where they’re going to college. All the things kids are supposed to be doing.”

SUPPORT DUKE CHILDREN’S

Complex treatments like Colby’s are made possible in part by your support. To make a gift to Duke Children’s, use the envelope in this newsletter or go to gifts.duke.edu/dch.
1. **Modell Diagnostic and Research Center Opens**

The Jeffrey Modell Diagnostic and Research Center was dedicated at a ceremony at Duke Children’s on Nov. 22, 2016. The Modell Foundation established the Center in partnership with CSL Behring. The Center, which will be directed by John Sleasman, MD, will offer advanced diagnostic evaluation to patients with a primary immunodeficiency. In addition to the generous initial gift to establish the center, the Modell family has pledged to match donations up to an additional $250,000 through March 1, 2018. Pictured here are (left to right): Sleasman, Fred and Vicki Modell, Mel Berger, MD, of CSL Behring, Ann Reed, MD, chair of the Department of Pediatrics, and Michael Herschfield, MD, Duke rheumatologist.

2. **Can’t Stop Dancing**

Congratulations and thank you to the student leaders who work all year to pull off dance marathons for Duke Children’s! The fourth annual Devilthon at Duke University raised $50,127. In its fifth year, Dance Marathon at NC State raised $77,000. UNC-Wilmington held its first Dance Marathon, called Dubthon, generating $32,534 with over 200 students participating. And Elonthon at Elon University (shown here) raised an incredible $351,554 for the kids in March!"

3. **Gamers for Good**

The fifth annual Extra Life gaming marathon was held in early November 2016. Over 650 gamers played video games for 24 hours and raised $110,259 for Duke Children’s – the largest amount to date. Special thanks to the Raleigh-Durham Guild which worked tirelessly to recruit gamers for Duke Children’s.

4. **Jump for the Children**

The 33rd annual Jump for the Children horse show to benefit Duke Children’s was held November 8-13, 2016, at the Governor James B. Hunt, Jr. Horse Complex in Raleigh. Spectators watched over 600 hunter and jumper horses compete throughout the week, culminating Saturday evening in the prestigious Grand Prix jumping event. Hosted by Triangle Farms, Jump for the Children is not just another horse show, but a horse show that makes a difference. To learn more about the 2017 show, visit jumpforthechildren.org.
YOU CAN BE A DUKE CHILDREN’S COMMUNITY PARTNER

Want to host your own fundraising event for Duke Children’s? **We love that idea!**

For information, please contact Amelia Howle at amelia.howle@duke.edu or 919-385-0033
Our community supporters put the FUN in fundraising!

5 Pediatric Research Retreat
The Duke Pediatrics Research Retreat is an annual event that offers the opportunity for students, postdoctoral fellows, clinical fellows, residents, research assistants, research associates, research scientists, and faculty members in Pediatrics to engage in collaborative discussions and showcase current research. The 2017 event was held April 18. Many of the life-changing research advances made at Duke Children’s are only possible through the support of generous donors like you!

6 IHOP, You Hop
The Duke Children’s Development team (above) and two Miss North Carolina local titleholders (below) enjoyed pancakes at IHOP on March 7, National Pancake Day. Participating restaurants around the country gave away free short stacks in exchange for donations to Children’s Miracle Network Hospitals, including the 25 IHOPs in North Carolina that raised funds for Duke Children’s.

7 In the Spirit
A gift of $45,070 from Spirit of Children stores will fund a position on the Duke Children’s Child Life team. Child Life provides enrichment and activities for Duke Children’s patients and offers a measure of normalcy to the inpatient and outpatient environments in the hospital. Spirit of Children has supported the Child Life program at Duke Children’s every year since 2010.

8 A Springtime Tradition
Guests enjoyed dinner, dancing, and an auction at the fourteenth annual Anna’s Angels Gala on March 25, 2017, at Prestonwood Country Club in Cary. Speakers included Duke Football head coach David Cutcliffe and Megan Bomgaars from A&E’s Born This Way. Anna’s Angels raises funds to support Down syndrome research at Duke Children’s.
YOUR LEGACY CAN HELP SICK CHILDREN

You can make a gift that benefits children in the future and provides income to you now. An irrevocable gift of $10,000 or more to Duke Children’s will give you fixed lifetime income and an income tax charitable deduction.

It’s easy to establish a charitable gift annuity. We will do all the paperwork for you and manage the funds. Duke agrees to pay you a fixed amount each year for the rest of your life.

sample payment rates

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Sample payment rates based on a donation of $20,000. Rates do not change once the annuity is established.

TO LEARN MORE, CONTACT US AT 919-385-3114 OR JOSEPH.TYNAN@DUKE.EDU.