JOHNS HOPKINS CHILDREN'S CENTER

Pediatrician

FALL 2010

Writing Away Weight?

JOURNALING IS AS IMPORTANT AS DIETING AND EXERCISE IN THIS PEDIATRICIAN'S CHILD OBESITY PROGRAM.

Journaling, notes pediatrician Maria Sofia Avendano-Welch—with 12-year-old Hailee Smith and her mom, Trudy—fosters self-esteem and healthy choices.

This past summer, First Lady Michelle Obama held her first live video chat on childhood obesity. Noting that one in three children in the United States are either overweight or obese, she stressed the importance of better eating practices and exercise, with parents leading the way. Cut down on the TV, go dancing, remove processed foods from the kitchen—and don't focus so much on the bathroom scale.

About the same time some 150 miles to the east, Salisbury, Md., pediatrician **Maria Sofia Avendano-Welch** was very actively practicing what Michelle Obama was preaching – a whole-family approach to curbing childhood obesity. Meeting that afternoon with three overweight children and their parents and even grandparents, Avendano-Welch was following up on their latest progress not so much in reducing pounds but in achieving a healthy lifestyle. But her practice's formula for a childhood obesity program came from patients and families themselves, not the First Lady.

"I learn a lot from my patients by listening to them and what they need, and developing a healthy lifestyle is something they're interested in," says Avendano-Welch. "So together we try to focus on healthy habits, not the weight."

Is it easy? No. But unlike patients who feel inhibited by strict dieting programs and their focus on numbers, Avendano-Welch's less-stressed patients and parents feel ownership in pushing toward a healthier lifestyle. It's about living, not losing. Most patients have lost weight and those who haven't have not gained any, either.

"In the long run, the weight loss will come as a result of the new healthy habits they've formed," says Avendano-Welch. "If they don't lose weight, they don't have to dwell on it. You don't want them to feel frustrated or overwhelmed, so we focus on the positive and the fact that they try."

How do they try? Frankford, Del., 12-year-old Hailee Smith writes down everything she eats and each time she works out with her Wii Fitness program. In each Wii yoga session alone, she boasts, she burns 220 calories. Also, she keeps more than a food and exercise diary—each day, as suggested by Avendano-Welch, she also writes down in her journal a goal and an achievement that made her feel better.

"When I exercise I feel better, and by losing weight I'll feel healthier," the 7th grader says. "And feeling healthier makes me feel a lot better about myself."

That positive sense of self through journaling, Avendano-Welch notes, creates and maintains momentum to keep alive the pursuit of a healthy lifestyle. "If they foster self-esteem, if they like themselves, they'll be more inclined to do something for themselves," she says. "The drive is more from them and consequently more effective."

For 9-year-old Cade Nutter of Hebron, Md., creating a healthier lifestyle meant a culture change for the whole family. First went the sodas, fried chicken and fries, replaced by diet-drinks, grilled chicken and fruit cups. Wheat bread, rather than white bread, now fills the pantry.

"We took everything out of the kitchen and started over," says Cade's mom, Jonna Twilley. "We all struggle with our weight, so we'll all suffer," she

continued on page 3

George Dover, M.D.
Director,
Johns Hopkins Children's Center
Given Professor of Pediatrics

Screening Athletes for **Sickle Cell?**

Beginning this fall, in a program rolled out by the National Collegiate Athletic Association (NCAA), all Division I college athletes will undergo mandatory screening for the sickle cell trait. The goal—to prevent rare but often-lethal complications triggered by intense exercise in those who carry the genetic mutation yet don't have the disease—is laudable, but the program is ill-conceived and underlines the difficulty in performing *mandatory* genetic screening tests. Athletes may simply opt out of screening by signing a school waiver.

Although research has shown that the risk of sudden death during extreme exercise is between 10 to 30 percent higher among those who have the sickle cell trait, there are much more reasonable ways to prevent these complications than mass genetic screening. For example, when the armed forces noted this increased risk in sickle-trait recruits during basic training, they instituted safeguards to reduce the risks of extreme exercise for *all* troops.

The NCAA program is rife with other pitfalls, too. Will positive test results be followed by a second test to eliminate false positives? Who will explain to students who test positive the difference between actual disease and carrier status and the risks associated with each? These were the questions I, along with Vence Bonhaj, J.D., and Lawrence Brody, Ph.D., of the National Human Genome Research Institute, raised in our commentary for *The New England Journal of Medicine* (Sept 9, 2010). Genetic screening must include clear education before screening, follow-up definitive testing to eliminate false positives, post-test counseling, and measures to prevent discrimination based on positive test results or refusal to be screened. In this case, safer practice conditions for *all* college athletes to reduce or eliminate the risk of death is preferable to mass genetic screening. Also, as this initiative will likely pave the way for other mass screening programs among college athletes, we need to proceed with caution. ■

Elevated Amino Acids Lead to a Sweet Diagnosis

The patient, a 7-day-old newborn, looked as healthy as could be when he showed up at the Hopkins Children's ED. He had come into the world via a normal vaginal delivery at 2.75 kilograms, had no family history of metabolic or genetic disorders, and suffered no visible symptoms aside from losing a little weight and sleeping more than usual in the previous 24 hours. On exam, his vitals were normal with the exception of a slightly elevated respiratory rate; his neuorologic evaluation showed all the appropriate reflexes.

"He appeared well, easily arousable and consolable," said pediatric resident **Sarah Kachan-Liu**.

So why was he in the ED?

The answer was in his newborn screen, which showed abnormally high levels of valine and leucine, so-called branched chain amino acids (BCAAs). What did the lab results point to? Hint: something you find in Vermont.

"We were worried that this child had maple syrup urine disorder (MSUD)," Kachan-Liu said, "unless there was a false positive in the newborn screen."

There wasn't as follow-up tests at Hopkins revealed valine at 944, four times its normal level of 202, and isoleucine and leucine well above their mean levels. The numbers for these BCAAs, said pediatrician and geneticist **Hilary Vernon**, were indeed diagnostic for MSUD, an autosomal recessive disorder first identified by Johns Hopkins neurologist John Menkes. In 1954 he reported four infants from one family who had all succumbed to the disease, and who all shared a common symptom – urine that smelled like burnt sugar and maple syrup. Hence the hallmark sign and name of the disorder.

"Patients may have no abnormalities on routine clinical testing," said Vernon, "but they can have this sweet or syrup-like body and urine odor."

Though rare, affecting 1 in 120,000 to 500,000 people, MSUD can be lethal if undetected and untreated. The disruption in the metabolism of the BCAAs and the resulting buildup of ketoacids can result in irritability and poor feeding by age 2-3 days, deepening encephalopathy by age 4-5 days, and coma and central respiratory failure by age 7-10 days.

"The typical presentation is a normal pregnancy, birth and one to two weeks of life, followed by a progressive toxic encephalopathy with ketosis," Vernon said. "Some children may present later if they have less severe enzyme deficiencies, which is something to consider in children with episodic neurologic disease who have not been screened in the newborn period," Vernon added, noting that today every state in this country screens for MSUD.

The good news is the mainstay treatment, long-term dietary management providing enough BCAAs for protein accretion and growth but not enough to trigger toxic metabolite accumulation, is effective. Pediatricians do have to monitor families frequently for compliance. Just as crucial, Vernon added, is emergency care.

"When these patients get sick and start breaking down endogenous tissues they're freely and uncontrollably releasing these amino acids that are poison to their body," Vernon stressed. "Neurologic decompensation can occur very quickly and create irreversible consequences, so it's very important to control the situation as fast as it's happening."

In this case the infant was given formula without the problematic amino acids. The result? His BCAAs decreased to normal levels over 10 days prior to discharge. "A pretty significant decompensation," said Kachan-Liu, "was avoided." ■

Hilary Vernon, M.D., Ph.D.

Pediatric cardiologist Joel Brenner with patient Alex Holtschneider and mom Debbie Holtschneider.

When the Referral is Urgent

After her son Alex, 15, suffered a couple of fainting spells while running, Debbie Holtschneider took him to his pediatrician's office. The diagnosis and recommendation? Dehydration and drink lots of water. Alex did, but the fainting did not cease. So the Bel Air, Md., mom consulted with a second pediatrician who immediately referred Alex to Hopkins Children's to be fitted with a Holter monitor, a portable device designed to measure electrical activity of the heart. There the mother wanted to see a pediatric cardiologist, but the next open appointment was in 30 days. Concerned her son might have a serious heart problem, she asked if she could talk to someone sooner. She was handed a card with pediatric cardiologist **Joel Brenner's** office number. Five minutes after she pulled out of the Hopkins garage, she called and Brenner answered. "Turn around and come back right away," he said after hearing her description of Alex's symptoms.

"Her son was passing out while running, which is never a good thing," Brenner says. "It may require emergency care."

Brenner explains that in general fainting in children and adolescents is a benign event, but when it occurs during exercise a potentially fatal condition may be the underlying cause. "Dehydration may be wishful thinking," Brenner says. "Heart rhythm and function problems are more likely, and require immediate attention."

Indeed, Brenner's workup and follow-up tests showed that Alex's heart was unable to expand properly with increasing exercise, a condition later diagnosed as restrictive pericarditis. While the double-layered sac surrounding the heart is designed to allow the heart to expand for optimal function, Alex's pericardium was scarred and compressing the right ventricle, constricting blood flow. After pediatric cardiac surgeons removed it, Brenner notes, blood flowed freely into the pumping chamber, restoring the teen's venous pressure. Looking back, he notes that this young patient could have had a much different outcome.

"He had a problem that limited him but could have been much worse," Brenner says. "Not only was his heart at risk of damage due to his limited cardiac output, but he could have suffered a traumatic head injury as a result of fainting while running or walking up a flight of stairs. This was one situation where appropriate testing needed to be carried out urgently."

The parents agree and couldn't be happier with the results, noting that today Alex plays both high-school tennis and travel soccer.

"Alex had a very unusual condition, and I don't know how many doctors would have been able to figure it out," says Mark Holtschneider. "Dr. Brenner's skillful diagnosis meant Alex could return to a normal life instead of living a life of inactivity, fear of the unknown, and serious future medical problems."

"Dr. Brenner didn't have to see Alex right away, he could have deferred to the scheduling secretary," adds Debbie Holtschneider. "But his hands-on 'do it now' approach meant that Alex got the immediate attention he needed."

When such referrals are urgent or not, Brenner explains, is pretty much determined on a case-by-case basis. Noting that pediatric cardiology, like other specialty clinics at Hopkins Children's, leaves open emergent slots each day, Brenner says, "The message is we try to be available when the primary care physician or parents raise a concern. In cases like this, there are very few guidelines beyond common sense." ■

Writing Away Weight

(continued from page 1)

laughs.

"Mindful eating" can also help, notes Avendano-Welch: "I tell my patients to eat slowly and to be aware of when they're full. Experience the food, listen to your body."

Cade enjoys playing baseball and soccer, but too much video gaming translated into too little time exercising, so the Xbox 360 took a leave of absence, too.

"I can no longer put sugar on fries," says Cade. "But the hardest part is having the Xbox taken away. I love it so much."

Reflecting on Avendano-Welch's patient-education focus on the long-term consequences of childhood obesity, Cade adds, "It's important to eat healthy and exercise so you don't have to give yourself needles every day for diabetes."

Ariana Urbano followed a similar route. The family made food selection changes across the board, with a focus on more white meat and reduced "child-fist-sized" portions, along with more exercise time. The 9-year-old says she has found a connection with jump rope. But even more helpful, she adds, is the support of her extended family. In addition to Ariana's mother and father, a grandmother and an aunt showed up with her at her latest clinic visit.

"Family involvement is really important, especially for younger children," says Avendano-Welch. "At this age the kids still look up to the parents and they listen to what they say. They all want to solve the problem." ■

For more information e-mail Dr. Avendano-Welch at *msa1037@aol.com*.

Treating Aerodigestive Diseases

Sarah Kilareski with her mom, Carrie, and gastroenterologist Karla Au Yeung.

Sarah was a "blessing from God," says her mother, Carrie Kilareski. "She was full of life and joy, a healthy baby."

But soon after her birth in October 2006, that healthy baby began to gag and spit up a lot. Her pediatrician wasn't too concerned, says Kilareski, because Sarah was gaining weight. But at her 12-month well-baby check she dropped from the 60th percentile on her weight growth chart at 6 months to 0 percent. Her mother was stunned: "She was eating, but she was losing weight."

Over the next three months, Sarah underwent a series of tests at a gastroenterology clinic near her Woodbridge, Va., home. She had reflux, but there had to be more to it than that, Kilareski thought as she watched her daughter decline in weight and spirit: "It was heartbreaking to watch your 1-year-old daughter become skin and bones. The doctors started using words like 'malnourished' and 'failure to thrive.'"

A barium swallowing test revealed that Sarah was aspirating thin liquids. She was referred to an ear, nose and throat specialist who concluded that Sarah's swallowing problem was related to a developmental delay. Add starch or gel thickeners to Sarah's fluids, he said, to reduce her risk of aspiration.

Kilareski did just that and at 18 months Sarah was up to the 50th percentile on her weight growth chart. But another swallowing test six months later showed she was still aspirating. Again the ENT suggested a developmental delay. "He said she'll grow out of it," says Kilareski. But she didn't.

Searching for a second opinion, Kilareski learned that Hopkins Children's was opening a new clinic for the treatment of so-called aerodigestive diseases – complex conditions involving the airways and upper digestive tract. Rather than be passed from one specialist to the next, Sarah would be evaluated by a team of pediatric gastroenterologists, otolaryngologists, pulmonologists and speech-language pathologists who would bring collective insights to bear in the case.

"Without such a clinic, individual specialists order procedures and treat patients often with a lack of coordination and input from other specialists," says pediatric gastroenterologist **Karla Au Yeung**. "That can delay the diagnosis because patients are waiting for months to see all of the doctors. Here patients get us all at once."

"We send a full note to the pediatrician saying what we collectively think needs to happen," adds pediatric otolaryngologist **Margaret Skinner**. "A lot of these children have been bounced from doctor to doctor only to be told they need to see four more."

To get to the bottom of Sarah's aspiration-reflux problem, Au Yeung used an endoscope to examine her esophagus, and Skinner a bronchoscope to look at her airway around the larynx and voice box. They both saw what had been previously missed – a small laryngotracheoesophageal cleft, an extremely rare and often easy-to-miss midline opening between the larynx, the trachea and the esophagus. Skinner describes the defect as a notch in the wall between the esophagus and trachea, through which liquids can pass. In severe forms, the notch can extend all the way down to the lungs, making it potentially lethal.

"Small clefts are difficult to diagnose and not something you can see in the office," says Skinner. "Sometimes you'll see it in a swallowing study with contrast sneaking through the back wall, but usually you need operative laryngoscopy and bronchoscopy to catch it."

Within two weeks of diagnosis, Skinner repaired the cleft in the operating room, and this past spring Sarah, for the first time, passed a swallowing study.

Sarah's mother was beyond relieved: "For almost two years we had been told by doctors that they did not know and might never know why Sarah was aspirating, so it truly was a surprise to finally get some answers." ■

For more information, call 410-955-8769 or 410-955-1686.

A Novel Way **to Get Lipids Down**

The patient, a 19-year-old from Ellicott City, Md., had levels of LDL (low density lipoprotein)—the so-called "bad" cholesterol—so high that he had to undergo coronary artery bypass at 15 years of age. Following the surgery, doctors used medications to lower his LDL cholesterol from 600-700 mg/dL to 300 mg/dL, but that was as low as they could go. With no apparent treatment in sight, that meant the teen was still at risk of significant atherosclerosis and coronary artery disease at a young age. But then he and his family learned of a novel, alternative cholesterol-lowering treatment called LDL apheresis being offered to patients like him at the Johns Hopkins Lipid Clinic. He underwent the procedure and amazingly his LDL cholesterol had fallen to 73 mg/dL.

"The procedure went great, and I'm so glad it could be done," says the boy's mother. "Without the treatment he could have died from his condition."

The condition she referred to is familial hypercholesterolemia, a genetic defect passed down through families that makes the body unable to remove LDL cholesterol from the bloodstream. Most patients with high cholesterol levels can be treated with a combination of diet, exercise and drugs, Lipid Clinic Director **Peter Kwiterovich** explains, but some patients, particularly those with more extreme familial hypercholesterolemia, do not respond as well to drug therapies. For them, LDL apheresis may be the sole answer. ■

For more information, call 410-614-0972.

When Anxiety Meets Headache

"These kinds of psycho-social interventions we think will be very effective and either reduce the need for medicines or reduce the dosage necessary."

—GOLDA GINSBURG, PH.D.

So, what came first, the anxiety or the headache? Are the two conditions related? A relevant question for many children, yet one few pediatricians may ask about.

"Pediatricians often treat migraine in their practice, but they often don't think about a relationship with anxiety or depression," says **Golda Ginsburg**, associate professor in the division of child and adolescent psychiatry at Johns Hopkins.

Following up on epidemiologic studies showing that some children with chronic headache have higher rates of psychopathology, particularly anxiety and depression, Ginsburg is leading a one-year pilot study exploring that relationship and treatment alternatives for these patients. Medicines are often the first-line treatment for headache pain, she notes, though they bring side-effects and the risk of over-use. The exacerbating role of anxiety, she adds, has largely been ignored. In this study, 30 patients ages 7 to 17 with a history of chronic headaches and excessive anxiety will receive family-based cognitive behavior therapy (CBT) or relaxation therapy (RT) with biothermal feedback.

"For a variety of reasons, many parents don't want to medicate their children," says Ginsburg. "These kinds of psycho-social interventions we think will be very effective and either reduce the need for medicines or reduce the dosage necessary."

"There is evidence showing that both of those therapies are effective in reducing anxiety and headaches," adds co-investigator **Kelly Drake**, also of the division of child and adolescent psychiatry. "We're trying to synthesize what we find into one treatment."

Co-investigator **Eric Kossoff**, a pediatric neurologist at Hopkins, agrees: "It's great to have many options for children with chronic headaches, especially those who don't respond to medicines. This study offers a potential new treatment for those children and their families."

In the study, known as the Children's Headache and Anxiety Management Program (CHAMP) and funded by the Migraine Research Foundation, half the patients will receive eight weekly one-hour sessions of CBT, and the other half eight weekly one-hour sessions of RT. Patients may continue to follow their existing medication regimen for their migraines while participating in the study, but no additional medications will be added. Patients' headache and anxiety symptoms will be evaluated before, during and one month after the therapy, and family factors like home environment and parenting behaviors will also be evaluated. ■

For more information, call 410-955-8021, or e-mail *kdrake2@jhmi.edu*.

Research Briefs

Marfan, A "Look-Alike" Disorder, or Neither?

Hopkins researchers have compiled what they believe are reliable lists of tell-tale physical signs to help doctors recognize children with Marfan and Loeys-Dietz syndromes. Timely and early diagnosis of both genetic disorders can mean the difference between life and death, but some of the most common physical features are also found in people with neither of the syndromes, which can cause confusion. Published as two separate studies in the August issue of the *Journal of Bone and Joint Surgery*, the two lists enumerate physical features that in certain combinations are highly suggestive of either Marfan or Loeys-Dietz syndromes, connective tissue disorders similar in presentation but caused by different genetic glitches. Many of the signal features of these disorders involve the face, skull, joints and spine, making them easy to spot during a physical exam, but not always easy to sort out. "The beauty of our lists is that they require no fancy imaging tests and most of the signs are right there for the pediatricians and the orthopedic surgeons to see," says co-investigator **Paul Sponseller**, director of Orthopaedics at Hopkins Children's. "All they have to do is see the forest for the trees. The lists will help them do so."

For more information, look under "News & Events" at *www.hopkinschildrens.org*.

ED Chief Douglas Baker **Reaches Out to Referring Docs**

There's no question Hopkins Children's new Director of Emergency Medicine M. Douglas Baker, is setting his sights on building a premier emergency medicine service for the new Charlotte R. Bloomberg Children's Center scheduled to open in early 2012. That means recruiting new staff and developing national leaders in pediatric emergency medicine. But in his dual role as Vice President for Community Outreach, there's another high-priority goal in his vision – strengthening relationships with referring physicians.

"We're able to practice a much better brand of medicine when we have that strong link between the community pediatricians and the university faculty," says Baker. "But that requires real-time communication between hospital staff and primary-care physicians, especially when the child is a patient in the ED or the medical center at large."

Baker should know – he brings a solid track record in connecting community and hospital physicians at the University of Texas Southwestern Medical Center, where he served as director of medical services and interim chief of pediatric emergency medicine, and at Yale-New Haven Children's Hospital, where he was chief of pediatric emergency services. Baker's work at Yale with faculty and a tightly knit community of several hundred pediatricians, he notes, was successful through improving day-to-day communication but also setting realistic goals.

"If the communication between house staff and community pediatricians didn't happen, faculty felt it was their misstep, but there was also a reciprocal expectation that the primary-care physician would be available to discuss clinical issues and responsible for the follow up," Baker says. "Real-time discussions between the attending physician in the hospital and the child's primary-care physician in the community facilitated a higher quality of care for our patients."

Baker, whose claim to fame as a clinical investigator is his groundbreaking research on the management of fever in infants, says he enjoys mentoring staff and helping them develop their own research projects. The joy of being an administrator, he says, is in creating and building programs. His arrival at Hopkins Children's, he adds, brings him full circle as he was recruited out of fellowship training at Boston Children's in 1984 to become the first pediatric emergency medicine physician at Hopkins and one of the first such specialists in the country.

"I started building that service at Hopkins Children's back then and have now returned to help complete the job," Baker says. "Having the new children's hospital open in a year and a half is certainly icing on the cake." ■

"Real-time discussions between the attending physician in the hospital and the child's primary-care physician in the community facilitated a higher quality of care for our patients."

—M. DOUGLAS BAKER, M.D.

Pediatrician

***Pediatrician* is produced by the Johns Hopkins Children's Center Office of Communications and Public Affairs.**
Medical Editors: Cozumel Pruette, M.D., Kenneth Schuberth, M.D.
Director: Kim Martin
Editor: Gary Logan
Contributing Writers: Katerina Pesheva
Designer: Max Boam
Photographers: Gary Logan, Keith Weller
Distribution: Naomi Ball
Web site: *www.hopkinschildrens.org*

TO REACH HOPKINS CHILDREN'S Physician to Physician
To refer or check on the condition of a patient, or to consult with a Hopkins physician, call 410-955-9444 or 1-800-765-5447 24 hours a day, 7 days a week

To contact the Johns Hopkins Children's Center Office of Development, call 410-516-4545.

The Johns Hopkins Children's Center
Office of Communications and Public Affairs
100 N. Charles Street/Suite 200
Baltimore, MD 21201
410-516-4741

Non-Profit Org
U.S. Postage
PAID
Baltimore, MD
Permit No. 1608