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› July/August 2009

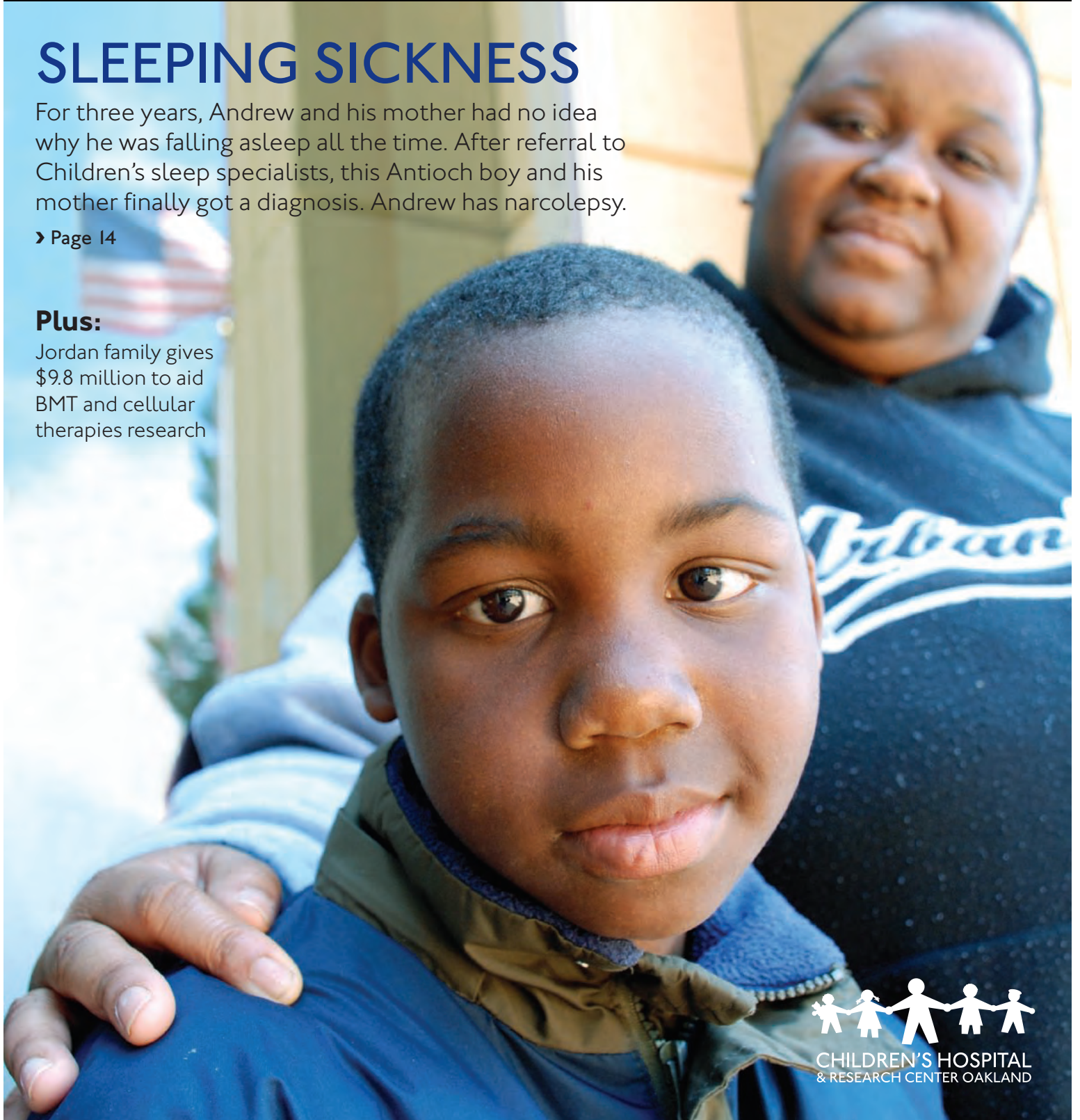
SLEEPING SICKNESS

For three years, Andrew and his mother had no idea why he was falling asleep all the time. After referral to Children's sleep specialists, this Antioch boy and his mother finally got a diagnosis. Andrew has narcolepsy.

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CHILDREN'S HOSPITAL
& RESEARCH CENTER OAKLAND

Sleeping sickness: Andrew wakes up

For three years, Andrew suffered mysterious bouts of sleepiness. Andrew's disorder, now diagnosed and treated by Children's subspecialists, is narcolepsy.

WRITTEN AND PHOTOGRAPHED BY TOM LEVY

When Andrew James, II, was 6, he regularly and unexpectedly began to fall asleep nearly every day—anywhere he happened to be—in the car, at school, while riding his bike or even while playing an exciting video game. For three years, physicians told Andrew's mother, Silvia, that he was fine. For three years, Andrew's unexplained sleepiness led teachers and a principal to call the boy lazy and his mother unfit.

But this was no ordinary sleepiness. In April 2009, after three years of fears and tears, subspecialists at Children's Hospital Oakland confirmed that Andrew, now 9, has narcolepsy.

Caused by a deficiency of hypocretin, also known as orexin, a brain chemical essential to sleep regulation, narcolepsy is more common in adults, but is also found in children. Sleep specialist Haramandeep Singh, MD, co-medical director of Children's Hospital's sleep lab, estimates that narcolepsy's prevalence is somewhere between 1 in 1,000 and 1 in 10,000 people. It's about the same as for multiple sclerosis.





Andrew was 6 when his kindergarten teacher first noticed he was sleeping a lot more than the other kids. That's when his mother, **Silvia**, first took the Antioch, Calif., boy to a local county clinic to find out what was wrong.

The physician there said Andrew was normal, that Andrew's fatigue, agitation and sleepiness would pass. "But it didn't go away like the doctor said it would," said Silvia.

Over the next three years, Silvia said teachers and the school principal said her son was lazy and called her an unfit mother. Her son was teased at school. Child Protective Services was called in.

"Andrew slept the entire year of first grade," said Silvia, the mother of five children. "He didn't learn anything. It got to the point where he didn't want to go to school."

Silvia began doing her own research into sleep disorders on the Web. She read up on narcolepsy and cataplexy. But when she mentioned these conditions during physician visits, she was told children don't have narcolepsy.

During their three-year odyssey, Andrew had four separate appointments at the county clinic, each with a different physician. And each time, except for the last, Silvia was told that Andrew was fine, that he was normal, that there was nothing wrong.

But the fourth visit was different; that's when the nightmare began to end. "Andrew started having symptoms while we were with the pediatrician," said Silvia.

At first, **Victoria Agnost, MD**, wasn't sure what was going on. "He was sitting in the office and he began to nod off," she said. Was it disordered nighttime sleep, or another problem that was causing the boy's sleepiness?

To Dr. Agnost, Silvia sounded like a very concerned mother trying to do the best she could. She didn't see parental neglect. But her instincts told her something was wrong.

"I tell my residents, nine times out of 10, when the parent has a concern, the parent is right," said Dr. Agnost. To her, the art of listening is as important as the art of medicine.

Dr. Agnost decided to refer Andrew to Children's Hospital.

Her decision lifted the weight of Andrew's thousand sleepy days off Silvia's back. "I'm very grateful to Dr. Agnost, because without her, we wouldn't even be here, knowing the diagnosis," said Silvia. "I would have gone crazy trying to figure out what was wrong with Andrew on my own."

At Children's Hospital, the first to see children suspected of sleep disorders are usually Pulmonary Medicine subspecialists. Andrew's first sleep-disorder appointment was March 3, with Pulmonary Medicine specialist **Danny Hsia, MD**. "Whenever you have a kid who has that much sleepiness, then you have to wonder if it's something more than lack of sleep," said Dr. Hsia.

Silvia told him about Andrew's sleepiness and Dr. Hsia asked whether there was evidence of cataplexy: losing muscle tone associated with emotions, especially laughter and anger. "Oh, he does that all the time," said Silvia. She also told Dr. Hsia about Andrew's occasional hallucinations of spiders and bugs when he was falling asleep or waking up.

It was enough for Dr. Hsia. He ordered a sleep study.

As psychiatrist, Dr. Singh began focusing on sleep



For more information about sleep disorders or to make a referral, call the Pulmonary Medicine department at 510-428-3305.

- Hiramandeeep Singh, MD, sleep lab director
- Danny Hsia, MD, Pulmonary Medicine

Hiramandeeep Singh, MD, co-medical director of Children's Hospital's sleep lab, became fascinated with sleep and sleep disorders while working as a psychiatrist in Minneapolis and Chicago.

"I was seeing sleep issues in the communities where I was working that weren't being treated," said Dr. Singh. "When I started treating them, these patients' quality of life improved incredibly. As a psychiatrist I was treating some symptoms,

such as depression, anxiety and cognitive issues, but I wasn't getting to the root of the problem. The gold is in sleep. There's a lot more that can be done if I'm treating sleep."

Dr. Singh sought more training, completing a Sleep Medicine Fellowship in July 2007 at the University of Texas Southwestern Medical Center in Dallas. He joined the Medical Staff at Children's Hospital Oakland in November 2007.

Dr. Singh had pre-

viously completed medical school at the Washington University School of Medicine in St. Louis. He did an internal medicine internship at the University of Chicago Hospitals and a psychiatry residency at Johns Hopkins Hospital in Baltimore. Dr. Singh practiced psychiatry for eight years before doing a sleep fellowship.

Dr. Singh is a board certified psychiatrist and board certified sleep medicine specialist.

Some science of narcolepsy and cataplexy

Few physicians receive training in the science of sleep. Dr. Singh, now a sleep specialist, said he had no exposure to sleep during medical school or residency. This makes it difficult for physicians to know when a sleep disorder may be the cause of a patient's symptoms.

The majority of patients missing the brain chemical hypocretin, also known as orexin, don't go to sleep; according to Dr. Singh, they are more likely to suffer periodic muscle tone loss. "As a result of not having sufficient hypocretin, REM sleep starts intruding into their wakefulness," he said, "so they'll have a burst of emotion and the part of REM causing muscle tone loss intrudes into their waking-state activity."

This can be very debilitating for many patients. They become extremely self-conscious, trying to avoid the emotional states, like laughter, fear and anger, that can trigger cataplexy.

Dr. Singh reports that research aimed at replacing hypocretin is ongoing. He also said that narcolepsy appears to have a genetic component; it runs in families. For example, if one parent has narcolepsy, the chance of their child having it is 1 in 4.

"Narcolepsy doesn't get diagnosed until way down the line because it gets miscategorized and misdiagnosed," said Dr. Singh. "A lot of patients out there are functioning and getting by with help from the medicines they've been getting, but they haven't gotten a formal diagnosis of narcolepsy."

Sleep disorder signs and symptoms

What pediatricians should look for when considering a sleep disorder, especially narcolepsy.

- Cognitive difficulties, including poor attention, distractibility, poor memory or diagnosis of ADHD
- Mood changes, including irritability, anxiety, depression and angry outbursts
- Excessive daytime sleepiness (falling asleep in class, long naps) or feels tired and lacks energy/motivation
- Automatic behaviors, including continuation of a behavior while falling asleep, for example: continuing to write nonsense on a piece of paper while falling asleep
- Hallucinations while falling asleep or when waking up
- Sleep paralysis (wakes up and cannot move, actual paralysis)
- Cataplexy, a sudden loss of muscle tone or weakness associated with laughter, anger or other strong emotions
- Poor school performance
- Frequent awakenings during sleep at night

National Institute of Neurological Disorders and Stroke
www.ninds.nih.gov/disorders/narcolepsy/detail_narcolepsy.htm



In a fifth-floor hallway in Children's Outpatient Center, Andrew shakes hands with Dr. Singh after a June follow-up appointment. His smiling mother, Silvia, is also pleased with the changes in Andrew's behavior and his ability to sleep better.

After an overnight polysomnogram to rule out other causes of sleepiness, such as significant sleep apnea, came the second part of the sleep study, a daytime series of four to five naps, with two-hour periods of wakefulness in between.

During this Multiple Sleep Latency Test (MSLT), measuring a child's daytime sleepiness, the child is in bed, with the lights out. REM sleep (dream sleep), normal at night, should not occur.

But the data gathered by electrodes measuring activity in his brain, muscles and heart showed Andrew was having REM sleep during his MSLT naps. "This kid was extremely sleepy," said Dr. Singh. "He fell asleep during every day and evening MSLT nap period and his average sleep latency—time to go to sleep—was about two minutes. Normal is in the 10-minute range."

Andrew met the diagnostic criteria for narcolepsy.

Hearing the diagnosis was a moment of vindication. "Silvia started cheering and crying," said Dr. Singh.

"She had been told by every single person in the school that this kid's lazy, that there's nothing wrong with him, that she was an unfit mother. I reassured her that we could get him help, that we could help him feel better."

Silvia finally felt a ray of sunshine. "I always knew there was something wrong with my baby," she said.

She relayed the diagnosis to her son's teacher and school principal. First they were incredulous, but Silvia said they now believe her and have been given Dr. Singh's phone number, as well as his note indicating the diagnosis and treatment.

Dr. Singh has prescribed interim stimulant medications, which Silvia gives Andrew twice a day to help promote daytime wakefulness. But Dr. Singh hopes to soon begin treatment with Xyrem, the preferred medication for treating daytime sleepiness and cataplexy together. It's the only FDA-approved medication for adults that treats both these symptoms of narcolepsy. It's also used to treat children, as an off-label medication.

Now Andrew can put a name to his disorder. "Now he can tell people it's narcolepsy," said Silvia. "It's not his fault."

After her three-year journey through a wilderness of brush-offs and accusations, Silvia has advice for physicians. "A lot of doctors need to listen more to their patients," said Silvia. "Don't be afraid to ask patients questions, a lot of questions. If they'd asked me questions, I could have told them a whole lot more."

But Silvia has only praise for Children's Hospital. "At Children's, they answer all my questions," she said. "I've been less stressed and more confident with Children's Hospital than with any other clinic."

With medication controlling his illness, Andrew's life has woken up. He's enjoying his favorite sports: soccer, basketball and tetherball. He's also catching up on the three years of learning he slept through. "I want to learn math and my multiplication tables," said Andrew. "I'm going to stay up more and do what everybody else does."