

The Washington Post

Mother labored to find reason for son's developmental delays

By Sandra G. Boodman
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Adults who encountered Adam Driscoll as a baby tended to be impressed by his quiet, easygoing nature. He seemed happy, cried little and, by the age of 6 weeks, even slept through the night. But his mother, Jen, though grateful for Adam's placidity -- a marked contrast to his rambunctious older brother -- viewed his undemanding behavior with an uneasiness that grew as he did.

A former day-care provider who worked in a genetics lab at the University of Delaware, Driscoll knew that siblings' personalities and development varied considerably. But she worried that Adam's behavior reflected something more ominous: His muscle tone was poor, and he was stumped by things that seemed almost instinctive to other children, such as playing with toys.

Driscoll repeatedly mentioned her concerns to her pediatrician, who brushed them off. "She told me, 'He'll be fine.' "

It was years before Driscoll, who encountered similar reactions from other specialists, learned the real reason for her son's difficulties, which by then were evident in many aspects of his life.

"He's very typical in that he presented symptoms as a newborn" but a diagnosis was

not made for several years, said Carole Samango-Sprouse, an associate clinical professor of pediatrics at George Washington University who is evaluating Adam, now 7.

"This is a disorder that can be easily identified or ruled out" but too often isn't, she said, because doctors are unfamiliar with the diagnosis or because they worry about labeling a child. As a result, the problem may go untreated for years, wreaking havoc on families who struggle to figure out what is wrong.

Adam was born four weeks early, and doctors told Driscoll and her husband, Toby, a math professor, that was the reason he had trouble feeding as a newborn. He seemed unable to suck but then learned to do it, his mother recalled, noting a recurring pattern. "From early on it seemed like we had to teach him," Driscoll said, recalling how she taught him to

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pick up blocks and use a shape-sorting toy, grasping his hand and guiding it. His limbs seemed somewhat floppy, but doctors ruled out cerebral palsy.

His experienced pediatrician dismissed Driscoll's increasingly urgent questions about Adam's development. "It's not like he won't be walking and talking when he goes to college," she recalled the doctor telling her.

But by 15 months, the age at which her older son walked, Adam showed no sign of taking steps. He wasn't talking or even babbling. The staffers at his day-care center were also concerned, and the pediatrician referred him to an early intervention program.

A nurse came to the Driscolls' home to observe Adam. To his parents' dismay, she pronounced Adam developmentally on track and said he was "just about ready to walk"; in fact, he did not take his first steps for eight more months, when he was nearly 2.

Social problems were also emerging. In day care Adam "hated to have other kids approach him. He wanted to be alone," his mother said. And he seemed terrified by swings or the slide.

When he was nearly 2 1/2 his family attended a Christmas party at the home of a developmental pediatrician with whom Driscoll worked. The doctor later told Driscoll that she was struck by Adam's behavior: First he hid behind the sofa and had to be coaxed

out. Then he sat quietly all night beside his parents, never running around and playing like the other children.

At Driscoll's request, the developmental pediatrician evaluated Adam, ruling out autism. He was given a diagnosis of an unspecified motor and speech delay and enrolled in a special-ed day-care center.

There he made significant gains, but by then Driscoll had become convinced something more global was wrong. "I felt that everything going on had to be tied together," she said. "There were too many problems in too many domains." Specialists working with Adam and his regular pediatrician counseled patience.

But a physical therapist agreed with Driscoll and urged her to see S. Charles Bean, a pediatric neurologist at the Nemours/Alfred I. duPont Hospital for Children in Wilmington, where Driscoll was then employed. "If

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anybody can figure it out, it's Chuck Bean," the physical therapist told Driscoll.

During an intensive, 90-minute evaluation in November 2007, Bean took a detailed history, watched Adam walk and eat, and played games with him. Then he ordered blood tests for several metabolic and genetic disorders. He told Driscoll he suspected Adam had fragile X syndrome, the most common cause of inherited mental impairment, which is responsible for a range of problems, from learning disabilities to mental retardation.

Driscoll thought fragile X was less likely than a mild form of muscular dystrophy.

Two weeks later, the results came back from the lab. "You're not going to believe this one," she recalled Bean telling her. They were both wrong.

Adam had Klinefelter syndrome, also known as XXY syndrome. The disorder, which is not hereditary, occurs very early in fetal development when boys randomly acquire an extra X sex chromosome rather than one X and one Y. Named for Harry Klinefelter, the physician who first described it in 1942, the syndrome affects one in 500 to 1,000 males, according to the Genetics Home Reference. In addition to being one of the most common chromosomal disorders, it is also one of the most stigmatized; early studies were conducted among prison inmates, and researchers erroneously linked the disorder with criminal behavior and low IQ, a legacy

that has been hard to shake.

"Early on it got a very bad press," said Samango-Sprouse, a neurodevelopmental specialist who works with children who have sex chromosome disorders. That linkage, she suspects, prevents some doctors from considering it as a possible diagnosis in children.

XXY syndrome varies in severity, although it almost always causes infertility because it impairs the production of testosterone. Males who do not have the extra X chromosome on all their cells may have mild cases and never know they have the problem until it is discovered during an infertility workup. Boys who are more severely affected may struggle with physical, social and language development. As babies, they tend to be shy and undemanding.

If the disorder is diagnosed early, boys can be treated with supplemental testosterone

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during puberty, which can boost levels of the male sex hormone during a critical growth phase, fueling the development of bigger muscles and more facial and body hair, which tends to be sparse. As teenagers, they may experience breast development; most are tall.

Bean said that the diagnosis can be difficult to make before puberty, because Klinefelter's mimics other problems. "It's not something I see frequently" as a neurologist, he said, unless a child has significant motor problems.

Driscoll said her initial reaction after Adam's diagnosis was to breathe "a huge sigh of relief. It explained everything and it's not going to kill him. I realized the path was pretty clear" about what needed to be done.

"But," she added, "it was a bit of a punch in the gut, too," because the diagnosis will affect Adam for the rest of his life. Driscoll said her family also grapples with the stigma that accompanies a genetic disorder, "especially one involving sex chromosomes," and with misinformation. When people learn that Adam possesses two X chromosomes, they often ask his mother if he is gay. (Studies have found that Klinefelter's patients are no more likely to be gay than the general population.)

Heart defects often accompany XXY syndrome, but a cardiologist found no such problems in Adam. He is continuing to r

ceive physical therapy and testing showed his intelligence is above average.

The Driscolls have told Adam about his diagnosis, explaining that he has an extra chromosome, "like an instruction book that was printed twice." She doesn't want Adam to think the problems are his fault, as did some men she has met whose disorder was not diagnosed until adulthood but had been told as children that they were lazy, dumb or mentally ill.

Adam, she says, is persistent and determined to surmount his difficulties. A few weeks ago, the little boy who used to hate playing with other children invited 10 classmates to his first-ever party to celebrate his seventh birthday.

Had Adam's condition been diagnosed earlier, Driscoll believes her family would have been spared years of unnecessary anxiety and frustration. "We feel tremendously lucky that

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we've gotten him diagnosed this young," she said. "But I worry about the other kids" whose baffling problems could be explained by a blood test.

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