

How autism in girls may help reveal the disorder's secrets

October 25 2016, by Lindsey Tanner



In this July 7, 2016, photo, Evee Bak, left, talks with her brother Tom Bak while they wait to talk with reporters at the Seaver Autism Center at Mount Sinai Hospital in New York. Tom has autism but Evee doesn't. The gender effect is a hot topic in autism research and one that could lead to new ways of diagnosing and treating the condition. (AP Photo/Seth Wenig)

Think autism and an image of an awkward boy typically emerges, but the way autism strikes girls—or doesn't—may help reveal some of the

developmental disorder's frustrating secrets.

Autism is at least four times more common in boys, but scientists taking a closer look are finding some gender-based surprises: Many [girls](#) with autism have [social skills](#) that can mask the condition. And some girls do not show symptoms of autism even when they have the same [genetic mutations](#) seen in boys with the condition.

"Autism may not be the same thing in boys and girls," said Kevin Pelphrey, an autism researcher at George Washington University.

The causes of autism aren't known. Genetic mutations are thought to play a role, and outside factors including older parents and premature birth also have been implicated. But the gender effect is now a hot topic in [autism research](#) and one that could lead to new ways of diagnosing and treating a condition that affects at least 1 in 68 U.S. children.

WHAT SCIENCE SHOWS

Brain imaging suggests there may be an additional explanation for why many girls with autism have more subtle symptoms than boys, Pelphrey said. Even in girls who clearly have autism, he said, brain regions involved in social behavior that are normally affected are less severely impaired.

Also, recent studies on autism-linked genes have found that girls can have the same kinds of genetic mutations seen in boys with autism, but not show symptoms. They "even need to have twice as many mutations on average to actually manifest with autism," said Joseph Buxbaum, director of an autism center at Mount Sinai medical school in New York.



In this July 7, 2016, photo, Erin Lopes gives a saliva sample for genetic testing while her daughter Eevee Bak looks on at the Seaver Autism Center at Mount Sinai Hospital in New York. Lopes' son Tommy was diagnosed at age 3 with autism. (AP Photo/Seth Wenig)

He is among researchers trying to identify a "protective factor" that may explain how some girls at genetic risk remain unaffected— perhaps a protein or other biological marker that could be turned into a drug or other therapy to treat or even prevent autism.

That possibility is likely a long way off, but Pelphrey said this line of research has prompted excitement among autism scientists.

AUTISM SISTERS PROJECT

Buxbaum is involved in the Autism Sisters Project, which is seeking to

enroll hundreds of families with autistic sons but unaffected daughters. The project began last year with the goal of building a big database that scientists can use to look for genetic clues and protective factors. Girls and their families visit the New York lab to give saliva samples for DNA analysis and efforts are underway to expand DNA collection to other sites.

Evee Bak, 15, hopes her samples will eventually benefit her older brother Tommy. The suburban Philadelphia siblings are just a year apart. They play in a garage band— Evee on drums, Tommy on guitar and vocals. He's a masterful musician, but has trouble reading social cues and doing things that come easy to other teens, like shopping alone or using public transportation.

Her focus is "taking care of Tommy and making sure he's happy and healthy," Evee said.

Tommy was diagnosed at age 3, after he stopped using words he'd learned months earlier and showed unusual behavior including repetitively lining up toys instead of playing with them.



In this July 7, 2016, photo, Eevee Bak gives a saliva sample for genetic testing while her mother, Erin Lopes, looks on at the Seaver Autism Center at Mount Sinai in New York. Scientists taking a closer look at autism are finding some gender-based surprises: Many girls with autism have social skills that can mask the condition. And some girls are born without autism despite strong genetic risks. Eevee, who doesn't have autism, hopes her saliva samples will eventually benefit her older brother Tommy, who has autism. (AP Photo/Seth Wenig)

"He's a wonderful person and I don't think that we'd ever want to change him," said his mother, Erin Lopes. But they'd welcome anything that could help him function as independently as possible "because I think that's what he really wants, is to be independent."

MAKING A DIAGNOSIS

Autism is diagnosed by observing behavior, there's no blood test for it.

Some experts say gender-based differences highlight a need to develop different ways to evaluate boys and girls.

Autism screening, recommended for kids starting at 18 months, uses tools based on research in autistic boys, said Rachel Loftin, clinical director of an autism center at Rush University Medical Center in Chicago.

One widely used screening questionnaire for parents includes questions like "Does your child play make-believe, make eye contact, seek praise, show interest in other children?" Girls with autism, especially mild cases, often don't show obvious problems in those categories—they're more likely than affected boys to play pretend with toys rather than lining them up by size or shape. Loftin said they're also more likely to show concern for another person's feelings.

Government data show that all forms of autism, mild to severe, are more common in boys and that the average age at diagnosis is 4 years in boys and girls. But Loftin said anecdotal evidence suggests a two-year lag time in diagnosis for girls, especially those with mild cases. And she suspects many cases are missed or misdiagnosed. That means a delay in early intensive behavior therapy that is the main treatment for autism.

Some girls manage to camouflage symptoms until pressures to fit in at school become overwhelming, delaying diagnosis until around age 8 or 9, said Alycia Halladay, chief science officer at the Autism Science Foundation, a nonprofit educational and research-funding group which is paying for the Sisters Project.



In this July 7, 2016, photo, Evee Bak, left, talks with her brother Tom Bak, center, while their mother, Erin Lopes, looks on at the Seaver Autism Center at Mount Sinai Hospital in New York. Autism is at least four times more common in boys, and Tom is affected, while Evee isn't. The gender effect is a hot topic in autism research and one that could lead to new ways of diagnosing and treating the condition. (AP Photo/Seth Wenig)

The prominent autism advocate, professor and author Temple Grandin wasn't fully verbal until age 4. "It was obvious something was drastically wrong with me," Grandin said. But she said she learned to adapt, in part because with "1950's parenting" she was faced with intense encouragement to develop social skills and other talents.

PARENTS' CONCERNS

Allison Klein worried about her daughter, Jillian, for three years before

the little girl was finally diagnosed with mild autism. Jillian couldn't tolerate loud noises, she grew withdrawn around her preschool classmates and she lagged behind academically. She was labeled anxious, not autistic.

"She didn't meet the stereotypical behaviors of no eye contact, no communication, hand flapping," Klein said. Teachers and doctors suggested she was just shy and would grow out of it.

A few months ago, just before Jillian turned 6, Loftin confirmed Klein's concerns.

Even Pelphrey, the [autism](#) researcher, had a similar experience. His daughter, Frances, was diagnosed almost four years after her behavior raised concerns. She didn't walk or talk until she was almost 3 years old. She tried to be "cuddly" and interact with others, but sometimes she did so awkwardly.

"Nobody really wanted to make the call," Pelphrey said. "Had she been a boy, there would have been much more pressure to look into it."



In this July 7, 2016, photo, Evee Bak, left, talks with her brother Tom Bak, center, and father, Tim Bak, while they wait to give saliva samples for genetic testing at the Seaver Autism Center at Mount Sinai Hospital in New York. Tommy was diagnosed at age 3 with autism, after he stopped using words he'd learned months earlier and showed unusual behavior including repetitively lining up toys instead of playing with them. Evee Bak, who doesn't have autism, hopes her saliva samples will eventually benefit her older brother. (AP Photo/Seth Wenig)

More information: CDC & Autism: tinyurl.com/zarzn2

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