

Gene deletion might be at core of autism

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Updated: October 17, 2011 - 8:38 AM



A cluster of genes is missing in children with autism, scientists at New York's Cold Spring Harbor Laboratory have discovered, a finding they say moved them a significant step toward unmasking the genetic underpinnings of the condition.

The subject of autism-related genes remains controversial in some quarters where parent groups still insist factors such as vaccines and toxins are at the core of the developmental disorder. But scientists at the lab have been on the trail of a suspect gene cluster since 2007, when geneticist Dr. Michael Wigler first proposed it may play a major role. Wigler suggested the missing cluster is a 27-gene grouping on chromosome 16.

Engineering autism mouse

Most people have two sets of the cluster; individuals with autism have only one, or just fragments of the second, researchers say. Now, Wigler's colleague, Dr. Alea Mills, has proved the deleted gene cluster not only plays a role in the condition, but also may affect head size, certain behaviors and the shape of structures within the brain itself. Mills said she chose to hunt down the cluster's role because "the idea that this deletion might be causing autism was exciting."

She still has to definitively prove the missing sequence has a hand in causing the condition.

The Centers for Disease Control and Prevention estimates one in every 110 children has autism, most of them boys. Mills proved the missing cluster is involved in autism by engineering a so-called autism mouse. "First we had to ask whether autism is such a human thing that we might be unable to model it in mice," Mills said.

When she clipped the corresponding cluster of genes from the mouse's genome, Mills discovered the animals developed hyperactivity and repetitive behaviors, two traits shared with humans who have autism.

"Mice with the deletion acted completely different from normal mice," said Guy Horev, a researcher in Mills' lab.

A link to head size

They found the deletion caused changes in the architectural shape of the animals' brain structures. Mills theorizes the suspect cluster also may be related to numerous features associated with autism in humans.

"Head circumference had been linked to autism in the past," she said. "Kids with autism tend to have larger heads. So larger head size is not only a predictor of autism, head circumference appears to be associated with the deletion."

Her research was funded by the Simons Foundation's Autism Research Initiative, a Manhattan philanthropic effort started by billionaire James Simons and his wife, Marilyn. Their daughter has Asperger's syndrome, an autism spectrum disorder.

Dr. Gerald Fischbach of the research center said: "The technical skill is extraordinary in creating mouse models bearing a human genetic variant ... associated with autism."

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