

Study uncovers why autism is more common in males

February 27 2014

Males are at greater risk for neurodevelopmental disorders, such as autism spectrum disorder (ASD), than females, but the underlying reasons have been unclear. A large cohort study published by Cell Press on February 27th in the *American Journal of Human Genetics* provides compelling evidence in support of the "female protective model," which proposes that females require more extreme genetic mutations than do males to push them over the diagnostic threshold for neurodevelopmental disorders.

"This is the first study that convincingly demonstrates a difference at the molecular level between boys and girls referred to the clinic for a developmental disability," says study author Sébastien Jacquemont of the University Hospital of Lausanne. "The study suggests that there is a different level of robustness in [brain development](#), and females seem to have a clear advantage."

A gender bias in the prevalence of neurodevelopmental disorders has been reported for ASD, intellectual disability, and attention deficit hyperactivity disorder. Some researchers have suggested that there is a social bias that increases the likelihood of diagnosis in males, whereas others have proposed that there are sex-based differences in genetic susceptibility. However, past studies investigating biological explanations for the gender bias have produced inconclusive results.

To examine this question, Jacquemont teamed up with Evan Eichler of the University of Washington School of Medicine to analyze DNA

samples and sequencing data sets of one cohort consisting of nearly 16,000 individuals with neurodevelopmental disorders and another cohort consisting of about 800 families affected by ASD. The researchers analyzed both copy-number variants (CNVs)—individual variations in the number of copies of a particular gene—and single-nucleotide variants (SNVs)—DNA sequence variations affecting a single nucleotide.

They found that females diagnosed with a [neurodevelopmental disorder](#) or ASD had a greater number of harmful CNVs than did males diagnosed with the same disorder. Moreover, females diagnosed with ASD had a greater number of harmful SNVs than did males with ASD. These findings suggest that the female brain requires more extreme genetic alterations than does the male brain to produce symptoms of ASD or neurodevelopmental disorders. The results also take the focus off the X chromosome for the genetic basis of the gender bias, suggesting that the burden difference is genome wide.

"Overall, females function a lot better than males with a similar mutation affecting brain development," Jacquemont says. "Our findings may lead to the development of more sensitive, gender-specific approaches for the diagnostic screening of neurodevelopmental disorders."

More information: A Higher Mutational Burden in Females Supports a "Female Protective Model" in Neurodevelopmental Disorders. *The American Journal of Human Genetics*, 2014; 94 (3): 415 DOI: 10.1016/j.ajhg.2014.02.001 . [dx.doi.org/10.1016/j.ajhg.2014.02.001](https://doi.org/10.1016/j.ajhg.2014.02.001)

Provided by Cell Press

Citation: Study uncovers why autism is more common in males (2014, February 27) retrieved 3

July 2023 from <https://medicalxpress.com/news/2014-02-uncovers-autism-common-males.html>

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