

## New study of autism reveals a 'DNA tag' (methylation) amenable to treatment

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A new discovery raises hope that autism may be more easily diagnosed and that its effects may be more reversible than previously thought. In a new study appearing online in *The FASEB Journal*, scientists have identified a way to detect the disorder using blood and have discovered that drugs which affect the methylation state ("DNA tagging") of genes could reverse autism's effects. This type of drug is already being used in some cancer treatments.

"As the mother of a now 22-year-old son with an [autism spectrum disorder](#), I hope that our studies as well as those of others, will lead to therapies that are designed to address specific deficiencies that are caused by autism, thus improving the lives of affected individuals," said Valerie W. Hu, Ph.D., one of the researchers involved in the work from the Department of Biochemistry and Molecular Biology at The George Washington University Medical Center in Washington, D.C. "Since autism is very diverse in the array of symptoms present in any given individual, it is first necessary to be able to identify specific deficits in each individual in order to design and then prescribe the best treatment. As an example of this personalized approach to medicine, we identified RORA as one of the [genes](#) that was altered specifically in the sub group of autistic individuals who exhibited severe language deficits."

To make their discovery, Hu and colleagues identified chemical changes in DNA taken from cells of identical twins and sibling pairs, in which only one of the twins or siblings was diagnosed with autism. The researchers then compared genes that showed changes in DNA tagging

(methylation) with a list of genes that showed different levels of expression from these same individuals. Then the scientists studied the amount of protein product produced by two genes that appear on both lists in autistic and control regions of the cerebellum and [frontal cortex](#) of the [brain](#). They found that both proteins, as predicted by the observed increase in DNA tagging, were reduced in the autistic brain. This suggests that blocking the chemical tagging of these genes may reverse symptoms of the disorder and demonstrates the feasibility of using more easily accessible cells from blood (or other non-brain tissues) for diagnostic screening.

"For far too long, autism research has been side-tracked by the cranky notion that it's caused by the MMR vaccine," said Gerald Weissmann, M.D., Editor-in-Chief of The [FASEB Journal](#). "Studies like this, which define genetic and epigenetic changes in discrete subgroups of the autism spectrum, offer real hope that effective treatments and accurate diagnosis are closer at hand."

**More information:** AnhThu Nguyen, Tibor A. Rauch, Gerd P. Pfeifer, and Valerie W. Hu. Global methylation profiling of lymphoblastoid cell lines reveals epigenetic contributions to autism spectrum disorders and a novel autism candidate gene, RORA, whose protein product is reduced in autistic brain. *FASEB J.* [doi:10.1096/fj.10-154484](https://doi.org/10.1096/fj.10-154484)

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