

## **Paternal Sperm May Hold Clues to Autism**

### **A look at the second paper from the EARLI Study**

#### ***What is this study about?***

Most experts agree that genetics contribute to the risk of autism – but the ways in which genes influence autism risk are very complex. In this study involving sperm samples, instead of focusing on how the actual genetic code influences autism risk, investigators examined the presence of molecules that get attached to genes (called “epigenetic tags”) that help regulate the genes’ activity.

#### ***Who did the study?***

Four groups worked on the study: Johns Hopkins University, Kaiser Permanente Division of Research, University of California, Davis, and Drexel University. The National Institutes of Environmental Health Services and Autism Speaks funded the study.

#### ***What did the study find?***

Using samples from 44 EARLI fathers, researchers found that DNA from the sperm of men whose babies later showed signs of autism at 12 months had distinct patterns of epigenetic tags.

#### ***Why was the study done?***

“We wondered if we could learn what happens before someone gets autism,” says Andrew Feinberg, M.D., M.P.H., of Johns Hopkins University School of Medicine. In addition to being easier to sample than egg cells from women, sperm are more susceptible to environmental influences that could alter the epigenetic tags on their DNA. “If epigenetic changes are being passed from fathers to their children, we should be able to detect them in sperm,” says co-lead investigator Daniele Fallin, Ph.D.

#### ***How was the study done?***

Shortly after their baby was conceived, some fathers enrolled in EARLI agreed to provide a sperm sample. One year after the child was born, the baby was assessed for early signs of autism using the Autism Observation Scale for Infants (AOSI).

The researchers collected DNA from each sperm sample and looked for epigenetic tags at 450,000 different positions throughout the genome. They compared the likelihood of a tag being in a particular site with the AOSI scores of each child. They found 193 sites where the presence or absence of a tag was statistically related to the AOSI scores. When they looked at which genes were near the identified sites, they found that many of them were close to genes involved in developmental processes, especially neural development.

#### ***Does this study suggest recommendations to the public?***

No. There is currently no genetic or epigenetic test available to assess autism risk.

#### ***Where was the study published?***

This paper was published in April 2015 in the *International Journal of Epidemiology*. The title is “Paternal sperm DNA methylation associated with early signs of autism risk in an autism-enriched cohort.” The full paper is available at: <http://dx.doi.org/10.1093/ije/dyv028>

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This summary was adapted from a Johns Hopkins Medicine news release, which can be found at [http://www.hopkinsmedicine.org/news/media/releases/paternal\\_sperm\\_may\\_hold\\_clues\\_to\\_autism](http://www.hopkinsmedicine.org/news/media/releases/paternal_sperm_may_hold_clues_to_autism)