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ASF launches Autism Sisters Project to accelerate research into 'Female Protective Effect'

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The Autism Science Foundation (ASF), a not-for-profit organization dedicated to supporting and funding autism research, today announced the launch of the Autism Sisters Project, a new initiative that will give unaffected sisters of individuals with autism the opportunity to take an active role in accelerating research into the "Female Protective Effect." The goal is to build a large genetic database that researchers can use to explore this phenomenon and discover how the protective factor can be harnessed to help people with autism of both sexes.

For years, scientists have reported higher autism prevalence in males, but the reason for this gender discrepancy isn't fully understood. One potential explanation is the presence of a protective factor in females that may be genetic, epigenetic, environmental, or a combination of multiple factors. Research has shown that some females carry genetic deletions or duplications that are known causes of autism, yet these girls do not exhibit clinical symptoms of autism. Other studies have pointed to the presence of a higher genetic "load" for females to reach the autism threshold, compared to males. As a group, girls with autism tend to exhibit more severe symptoms and tend to be diagnosed later. These initial findings warrant a focused study of unaffected sisters of individuals with autism to try to identify this potential protective effect.

"We are learning more about how autism affects males and females differently, as well as the underlying etiological factors behind these differences," said Alycia Halladay, PhD, chief science officer of the Autism Science Foundation. "This is an exciting and promising opportunity to leverage that understanding for deeper research into potential factors that could have a significant impact on the lives of many people with autism. Right now, the limiting factor is a lack of genetic data. The Autism Sisters Project will help eliminate that barrier and move the science forward."

The Autism Sisters Project will focus on three areas:

- Data on unaffected sisters will be gathered from existing databases with rigorous behavioral phenotyping data on all family members, including unaffected siblings, beginning with samples in the Autism Sequencing Consortium;
- Funds will be provided to autism research sites so that sequencing and phenotyping can be expanded to include an unaffected sister in families where samples from parents and the individual diagnosed with autism have already been collected
- New families with a member who has autism and a female sibling without an ASD diagnosis will be recruited to the Icahn School of Medicine at Mount Sinai to donate saliva samples and participate in a full screening. A full DNA exome scan, among other analyses, will be performed on the entire family.

The project will convene a scientific advisory panel in November led by Joseph D. Buxbaum, PhD, Director of the Seaver Autism Center at the Icahn School of Medicine at Mount Sinai, along with experts in genetics, statistical genetics, epidemiology, and ASD clinicians. The panel will develop a study protocol that will allow the question of the female protective effect to be properly addressed. The other advisory panel members are: Somer Bishop, PhD (UCSF); Ed Cook, MD (University of Illinois at Chicago); Mark Daly, PhD (Harvard Medical School/Broad Institute); Bernie Devlin, PhD (University of Pittsburgh); Elise Robinson, PhD (Harvard Medical School); Kathryn Roeder, PhD (Carnegie Mellon); Stephan Sanders, PhD (UCSF); Celine Saulnier, PhD (Marcus Autism Center at Emory); Paige Siper, PhD (Mount Sinai); Huda Zoghbi, PhD (Baylor); and Lonnie Zwaigenbaum, MD (University of Alberta).

"The female protective effect is a very important area of investigation in the autism research community and the Autism Sisters Project is going to jumpstart the process of developing a necessary cohort of unaffected female siblings," said Dr. Buxbaum. "I, and all my colleagues at Mount Sinai, are thrilled to be partnering with the Autism Science Foundation on this initiative. This is an enormously exciting opportunity for sisters of individuals with autism to take a proactive role in advancing important research."

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