

FROM: **AUTISM SCIENCE FOUNDATION**

Rubenstein Communications, Inc.  
Adam Pockriss – 212.843.8286 [apockriss@rubenstein.com](mailto:apockriss@rubenstein.com)  
Julia Tomkins – 212.843.9223 [jtomkins@rubenstein.com](mailto:jtomkins@rubenstein.com)



---

**FOR IMMEDIATE RELEASE**

**THE AUTISM SCIENCE FOUNDATION LAUNCHES THE AUTISM SISTERS PROJECT,  
TO SEARCH FOR AUTISM'S "FEMALE PROTECTIVE EFFECT"**

***Sisters of Individuals with Autism Can Now Visit the Seaver Autism Center at The Mount Sinai Hospital  
in New York to Get Involved with the Research Effort***

NEW YORK, NY (October 13, 2015) – 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.”

To participate in the Autism Sisters Project, interested participants should contact the Seaver Autism Center by phone at 212-241-0961 or by email at [theseavercenter@mssm.edu](mailto:theseavercenter@mssm.edu).

#### **About the Autism Science Foundation**

The Autism Science Foundation (ASF) is a 501(c)(3) public charity. Its mission is to support autism research by providing funding to scientists and organizations conducting autism research. ASF also provides information about autism to the general public and serves to increase awareness of autism spectrum disorders and the needs of individuals and families affected by autism. To learn more about the Autism Science Foundation or to make a donation, visit [www.autismsciencefoundation.org](http://www.autismsciencefoundation.org).

#### **About the Seaver Autism Center for Research and Treatment at Mount Sinai**

The Seaver Autism Center for Research and Treatment at Mount Sinai conducts progressive research studies aimed at understanding the multiple causes of autism spectrum disorder (ASD). The multidisciplinary team is comprised of experts in the fields of genetics, molecular biology, model systems, neuroimaging, and experimental therapeutics who are dedicated to discovering the biological causes of ASD. The Center strives to develop innovative diagnostics and treatments for integration into the provision of personalized, comprehensive assessment and care for people with ASD. The Seaver Autism Center was founded through the generous support of the Beatrice and Samuel A. Seaver Foundation. For more information, visit [www.seaverautismcenter.org](http://www.seaverautismcenter.org).

#### **About the Autism Sequencing Consortium**

Founded in 2010 by Joseph D. Buxbaum, PhD, the Autism Sequencing Consortium (ASC) is an international group of scientists who share autism spectrum disorder (ASD) samples and genetic data. All shared data and analysis is hosted at the Icahn School of Medicine at Mount Sinai on a supercomputer called Minerva designed by Mount Sinai faculty, which enables joint analysis of large-scale data from many groups. The ASC is supported by a cooperative agreement grant to four lead sites

funded by the National Institute of Mental Health (NIMH), with additional support from the National Human Genome Research Institute (NHGRI). The PIs are Drs. Joseph D. Buxbaum (Icahn School of Medicine at Mount Sinai), Mark J. Daly (Broad Institute of MIT and Harvard), Bernie Devlin/Kathryn Roeder (University of Pittsburgh School of Medicine and Carnegie Mellon University), and Matthew State (University of California, San Francisco).

###