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AUTISM SCIENCE FOUNDATION ANNOUNCES
RECIPIENTS of SPRING 2017 PRE- AND POST-DOCTORAL FELLOWSHIPS

Nine Early Career Scientist and Mentor Teams Receive Funding

NEW YORK, NY (April 27, 2017) – The Autism Science Foundation, a not-for-profit organization dedicated to funding innovative autism research, today announced the recipients of its annual pre- and post-doctoral fellowship grants. Three pre-doctoral and six post-doctoral fellowship grants will be awarded to student and mentor teams conducting research in deep brain stimulation, gene and environmental interactions, epigenetics, pain response, neurobiology, and sex differences in ASD.

"Each of the projects selected for funding has the potential to improve the lives of people with autism," said Autism Science Foundation president Alison Singer. "We are pleased to support the work of this impressive group of young scientists and look forward to the progress that will be made as a result of their efforts."

The FRAXA Research Foundation, a non-profit that funds biomedical research to develop treatments for Fragile X Syndrome (FXS), a monogenic cause of autism, will co-fund the post-doctoral fellowship awarded to Carol Wilkinson, MD PhD, of Boston Children's Hospital. Wilkinson's research will examine biomarkers of individuals on the more severe end of the autism spectrum and study single-gene disorders associated with ASD, like FXS.

"FRAXA and ASF have collaborated for many years, which is appropriate considering the many links between fragile X and autism," said Dr. Michael Tranfaglia, medical director and chief scientific officer of the FRAXA Research Foundation. "We are excited about our latest joint project at Boston Children's Hospital, which will study human electrophysiology, with an eye toward future use as an outcome measure in clinical trials."

Since its founding in 2009, the Autism Science Foundation has funded more than $3 million in grants, including pre- and post-doctoral fellowships, medical school gap year research fellowships, three-year early career awards, treatment grants, undergraduate summer research grants, research enhancement mini-grants, and travel scholarships to enable stakeholders to attend the annual International Meeting for Autism Research (IMFAR).

The following pre- and post-doctoral projects were selected for 2017 funding:

Autism Science Foundation's Pre-Doctoral Fellowship Grant Recipients

Songjun William Li, Boston University School of Medicine
Advisor: Ziv Williams, MD
Exploring the possibility of deep brain stimulation in autism
Deep brain stimulation is a therapy used for many neurological disorders but so far has been not well understood in those with autism spectrum disorder. In order to better understand the specific areas for autism-related behaviors that would be the target of this therapy, this research group will study monkeys that are trained in a social interaction task. The researchers will look at how the cells work at different times during this task, and stimulate certain cells to better understand how deep brain stimulation may help people with ASD.

Elizabeth Sharer, University of Minnesota
Mentor: Jed Elison, PhD
Defining the female protective effect in infants with ASD
Four times as many boys as girls are diagnosed with autism, and multiple lines of evidence suggest a "female protective effect" as one explanation for the sex bias. Past research has focused primarily on females who have been diagnosed with ASD, but it is unknown whether the protective effect is evident in females who show some symptoms of autism but not enough to warrant a full diagnosis. Examining brain and behavioral development from toddlerhood, this is the first study to investigate the female protective effect in infants who show behaviors of concern, as compared to those who develop typically and those that are later diagnosed with ASD. This will help efforts to identify those with autism and other concerns as early as possible, and may provide more personalized approaches to treatments for females with autism.

Christina N. Vallianatos, University of Michigan
Mentor: Shigeki Iwase, PhD
Illustrating the importance of epigenetics in the sex bias of ASD
While the genetic influences in ASD have received attention, chemical modifications to DNA, or epigenetics, are less understood. By tagging certain parts of the DNA or altering its shape, environmental factors can change expression of genes related to ASD. In addition, epigenetics may play a role in why more males are diagnosed with ASD than females. This study will examine the function of a gene important in epigenetics called KDM5C. So far, this gene is associated with autism and intellectual disability in males, therefore research using animal models has been limited to males. Therefore, this study will look at a mutation of KDM5C in females, focusing on behavioral features, brain development and "downstream" gene expression. This research will shed light on the mechanisms of possible gene/environment interactions in a previously understudied group of people with autism: females.

Autism Science Foundation's Post-Doctoral Fellowship Grant Recipients

Michael S. Breen, PhD, Mount Sinai School of Medicine
Mentor: Joseph Buxbaum, PhD
Developing peripheral blood and neuronal biomarkers for autism using a genetically defined subtype
This project will begin to determine the potential validity of a blood based biomarker for autism by comparing gene expression in blood and IPSC cells from the same patients. In this project, Dr. Breen will use information from individuals with a severe form of autism characterized by a mutation of the SHANK3 gene. This work will begin to identify new genes/pathways in blood samples that will improve

diagnosis and also identify new drug targets that will enhance the development of new treatments. These findings can then be applied to a broader range of people with ASD.

Isabella Rodrigues Fernandes, PhD, University of California at San Diego
Mentor: Alysson Renato Muotri, PhD
Screening for new autism treatments using cells in a dish
The Muotri lab is using a cell culture system which more accurately models the complexity of the human brain compared to just single brain cells. Including other cells found in the brain, like astrocytes, allows researchers to identify additional factors that affect brain function. In this way, the researchers can recreate many biological features of ASD, incorporating networks of brain cells acting together and the inflammatory responses of astrocytes. Using this system, the researchers will screen 10 potential therapeutic drugs in a system where the cells have an autism-related mutation and examine not just the effect of neurons, but other brain cells as well. This will impact the way that molecules for treatment are identified, screened, and then moved into clinical trials.

Michelle Fialla, PhD, Vanderbilt University
Mentor: Carissa J. Cascio, PhD
Understanding the pain response in people with autism
Because of difficulties communicating, people with autism may not be able to express their response to physical pain in a variety of situations. It is also possible that sensory issues may change processing to pain, or the interpretation of stimuli as painful or not painful. Very little research has been done to address this crucial issue affecting those with ASD and impacting their clinical care plan. This study will examine verbal and non-verbal responses to a mild stimuli in adults with ASD. The findings will help clinicians understand pain sensitivity in those with autism so that new strategies to assess and manage pain can be developed.

Dorothea Floris, PhD, New York University Medical Center
Mentor: Adriana Di Martino, MD
Characterizing the female and male brain in autism
As a result of data sharing efforts, neuroimaging databases with thousands of pieces of data covering hundreds of individuals with and without autism now exist. Using two of these databases, this project will examine functional magnetic resonance imaging data (fMRI) to better understand which areas of the brain are over connected or under connected in autism. By combining databases of imaging data, the researchers will be able to include enough females to understand how brain connectivity in males and females with autism are similar and different. Ultimately, this will inform future development of gender and sex-specific diagnostic criteria and interventions.

Eitan Kaplan, PhD, Seattle Children's Research Institute
Advisor: Robert F. Hevner, PhD
Determining the genetic and environmental factors influencing brain development
Scientists agree that most cases of autism are the result of genetic and environmental interaction. In this study, researchers will explore how the development of new brain cells are affected by the combination of a genetic mutation in a known autism risk gene (TBR1) and the environmental factor of an immune response during pregnancy. This novel approach will better describe the role of each risk factor – genetic and environmental – separately, and together on the formation of the cerebral cortex, a brain region known to be involved in ASDs.

Carol Wilkinson, MD PhD, Boston Children's Hospital
Mentor: Charles A. Nelson, PhD
Developing biological markers for more severely affected individuals
There is great variability in cognitive function, language ability and behavioral symptoms in people with autism. Studying single-gene disorders associated with ASD, such as Fragile X Syndrome (FXS), can address the differences seen across the spectrum of autism. Nearly half of all children with FXS meet criteria for ASD, and virtually all have cognitive and language difficulties ranging from mild to severe. This provides an opportunity to examine biomarkers of individuals on the more severe end of the autism spectrum. This project will use non-invasive brain activity measures to understand what happens in the brains of young children with Fragile X during in response to sensory stimuli, and how this correlates with symptoms often seen in autism. These brain-based markers will then be used in future clinical trials as objective measures for targeted outcomes. This fellowship is supported through a partnership between the Autism Science Foundation and the FRAXA Research Foundation.

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.

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