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Molecular Autism Receives Highest Impact Factor for an Autism Journal

On July 29, 2014, Thomson Reuters awarded an Impact Factor of 5.486 to the open access journal *Molecular Autism*, co-edited by Joseph D. Buxbaum, PhD, Director of the Seaver Autism Center. This represents the highest Impact Factor for any journal dedicated to autism or related neurodevelopmental conditions.

The journal was created in 2010, by Dr. Buxbaum, also a Professor in the Departments of Psychiatry, Neuroscience, and Genetics and Genomic Sciences at the Icahn School of Medicine at Mount Sinai, and Simon Baron-Cohen, PhD, Director of the Autism Research

Centre at the University of Cambridge. The goal of the journal was to provide an outlet for the volume of exciting genetic and other molecular autism research papers, and to make this cutting-edge autism research available freely via open access. In the past four years, *Molecular Autism* has grown and now publishes approximately five articles per month.

Since its launch, *Molecular Autism* has published over 100 articles. The journal averages 30,000 accesses per month, and April 2014 saw a record



Molecular Autism
Brain, Cognition and Behavior

IMPACT
FACTOR
5.49

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Distinguished Lecturer Series

On September 10-11, 2014, the Seaver Autism Center hosted the Distinguished Lecturer Series (DLS). The DLS is unique because each installment includes one lecture geared towards researchers and health care professionals, and one prepared specifically for caregivers of people with autism and the general public. The goal of the DLS is to enhance knowledge for all attendees, and educate various groups within the autism community.

Bernie Devlin, PhD, Professor of Psychiatry and Human Genetics at the University of Pittsburgh School of Medicine, and Kathryn Roeder, PhD, Professor of Statistics and Computational Biology at Carnegie Mellon University, visited together and each gave one of the presentations as part of this event.

If you would like to be notified of similar events in the future, please email seavercentereditor@mssm.edu.



Dr. Devlin's presentation, titled "Genetics opens a window to view risk for autism," was tailored to a non-scientific audience. Immediately following was a reception for all guests to mingle and ask Dr. Devlin further questions.



After meeting with Mount Sinai researchers and trainees, Dr. Roeder gave a presentation titled "Network Assisted Analysis Helps Reveal the Genetic Basis of Autism" designed for a scientific audience.

Open Letter

As the mother and sister of a person with autism, and the founder and president of the Autism Science Foundation, I am proud and excited to be part of *Autism BrainNet* and the *It Takes Brains* public outreach program. *Autism BrainNet* is a new research initiative that brings together leading research institutions to collaborate on autism brain research, and *It Takes Brains* is intended to raise awareness of the severe shortage of brain tissue available for needed research and to urge people to register to become a brain tissue donor after death.

Autism BrainNet was formed because postmortem studies on brain tissue represent the best way for researchers to gain a deeper understanding of autism on the genetic, cellular, and molecular levels, and the most promising way by which scientific research can lead to improvements in the quality of life for those on the autism spectrum. If you have autism, if you are a family member of someone with autism, or even if you are unrelated to anyone with autism, you now have a unique opportunity to join families like mine who are registering with *Autism BrainNet* in order to contribute to a better future for people dealing with the challenges of autism.

The *It Takes Brains* website (www.takesbrains.org) provides detailed information about registration and donation. The people behind *Autism BrainNet* understand that brain donation is a difficult subject for many and takes courage to discuss, and the *It Takes Brains* website covers many issues of concern to potential donors and their families. You will learn, for example, that brain tissue donation does not involve any cost to the donor's family, is permitted by most religions, and does not alter funeral arrangements or medical treatment. Further, while

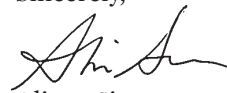
advance registration is encouraged, it is not required and it is not binding. Donor families will be treated with respect and compassion, and provided with continuing support and information.

Autism BrainNet is currently comprised of the Icahn School of Medicine at Mount Sinai in New York, the MIND Institute of the University of California at Davis, the University of Texas Southwestern Medical School, and the Harvard

University/Beth Israel Deaconess Medical Center. There are plans to add sites for brain research across the U.S. and internationally.

Please visit www.TakesBrains.org and www.autismbrainnet.org to learn more about brain research and to register.

Sincerely,



Alison Singer



THE MATTHEWS, FROM BEACON, NY; CHRISTINE, MARK AND THEIR SONS MARK JR., TIMOTHY AND CASEY.
CASEY, AGE 14, HAS AUTISM.

IT TAKES BRAINS TO SOLVE AUTISM

The Matthews never saw themselves as a "super" family, yet when they bonded together to support their autistic son Casey, they were indeed super. And now the Matthews and families like them around the world are being looked upon as super heroes. Why?

They have all been united by one brave and heroic act...pledging to donate the brain tissue of their sons and daughters for when they are sadly no longer with us. It's difficult to think about, but the reality is that brain tissue is urgently needed for the scientific research that will help thousands of people with autism.

Join forces with the Matthews and other super hero families to help ensure a brighter future for all. Because it takes brains to solve autism.

Visit TakesBrains.org to learn more and see how your entire family can help build a better tomorrow.



This initiative is guided by:



Molecular Autism

Continued from page 1

of over 40,000 accesses.

The Impact Factor (IF) is a measure of the frequency with which the “average article” in a journal has been cited in a particular year or period, and the annual Journal Citation Report (JCR) IF is a ratio between citations and recent citable items published. This IF is the first for *Molecular Autism*, after being tracked for several years, and it places the journal among the highest in the broader fields of neurosciences and genetics based on category information

from the 2013 JCR. It ranks 34 out of 251 journals in Neuroscience, and 21 out of 164 journals in Genetics & Heredity.

While impact factors are one way of measuring impact, the strong performance of *Molecular Autism* provides further evidence that open access journals are delivering high rates of citation as well as high visibility.

Dr. Buxbaum said, “The high Impact Factor reflects the significance and importance of the articles found in *Molecular Autism*, and having such a

high IF will ensure that we will continue to receive high-impact manuscripts describing key studies in autism and related conditions.”

Dr. Baron-Cohen added, “We are extremely proud to have launched this journal and achieved this milestone. We greatly appreciate the researchers that have submitted top-quality studies, the tireless reviewers, and the editorial staff from BioMed Central.”

For more information, visit www.molecularautism.com.

New Grant Awards

► Rare Disease Clinical Research Consortium (RDCRC) – “Developmental Synaptopathies Associated with TSC, PTEN, and SHANK3 Mutations” from the National Institute of Neurological Disorders and Stroke

- Alex Kolevzon, MD, Clinical Director of the Seaver Autism Center, will lead the Phelan-McDermid Syndrome longitudinal study as part of this RDCRC, and Joseph D. Buxbaum, PhD, Director of the Seaver Autism Center, will act as Administrative Director of the Consortium. This grant marks the first time the National Institutes of Health has provided any funding for the clinical and longitudinal assessment of Phel-

an-McDermid Syndrome (PMS)/SHANK3 mutations, which we now know to be a relatively common monogenic cause of autism. This study will comprehensively characterize the phenotype and natural history of PMS, identify biomarkers using neuroimaging, and identify genetic factors which contribute to diverse phenotypes in patients with PMS.

► “Population-Based Autism Genetics & Environment Study” from the National Institute of Mental Health

- Led by Dr. Buxbaum, this study will use a population-based epidemiological sample with detailed demographic and environmental information to assess the role of inherited and

de novo, or genetic, variants in autism. The researchers will also evaluate rare standing variation in autism, while integrating key environmental variables.

► “Prefrontal function in the Shank3-deficient rat: A first rat model for ASD” from the National Institute of Mental Health

- This study will be led by Drs. Buxbaum and Patrick R. Hof, Vice Chair for the Department of Neuroscience at the Icahn School of Medicine, and it will make use of the first-ever rat model of autism, created here at Mount Sinai by researchers

in the Seaver Autism Center. The researchers will conduct behavioral assessment, electrophysiological analysis, and neuropathological and neurochemical investigation of prefrontal function in Shank3-deficient rats.

► “Neural Effects of Sustained Oxytocin Treatment in Children with Autism” from the Eunice Kennedy Shriver National Institute of Child Health and Human Development

- Led by Ting Wang, PhD, Assistant Professor of Psychiatry in the Seaver Autism Center, this research will examine the impact of sustained oxytocin treatment on the social cen-

ters of the brain in children with autism spectrum disorders to better understand changes in the brain that predict and accompany response to treatment.

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• **SEAEVER IS CONTINUING TO GO GREEN!** Please send your email address to seavercentereditor@mssm.edu to receive this newsletter electronically.

Study Reveals Common Genetic Changes Are Significant in Autism

Genetic changes are responsible for roughly 60 percent of the risk for autism, and most of these variants are commonly found in the general population, according to a groundbreaking study led by Dr. Joseph Buxbaum.

The remaining nongenetic factors that account for roughly 40 percent of the risk for autism are not known. However, environmental factors and the interaction between genes and the environment may be a part of these nongenetic factors.

Findings from the four-year study—called the Population-Based Autism Genetics and Environment Study (PAGES) Consortium—were published in the July 20, 2014 issue of *Nature Genetics*.

In the study, the largest of its kind ever

undertaken, Mount Sinai researchers worked in collaboration with Swedish scientists, analyzing 500,000 common genetic variants from 3,046 people, 466 of whom had autism. The team then corroborated their results with a longitudinal Swedish study that followed more than 1.6 million families, and included more than 14,000 individuals with autism.

“Most of the risk for autism is found in common variants, but we know that in a specific individual, a spontaneous mutation or other rare mutation is often the decider, with the interaction between the common variants and rare mutations leading to the diagnosis,” Dr. Buxbaum says. “There was already evidence two years ago that both common and rare genetic variation contributed to autism. Using this unique sample, we were able to make it perfectly clear regarding

relative contributions, and showed that common variation is significantly more important than generally appreciated.” The study was funded by the National Institutes of Health (NIH) and the Beatrice & Samuel A. Seaver Foundation.

Dr. Buxbaum is currently leading a multinational consortium that is completing a study of 3,800 individuals with autism to identify spontaneous and other rare mutations as well. The consortium is using advanced methods to look at every gene in the genome.

“Together, with support from the NIH and enormous philanthropy from the Seaver Foundation, and others, we are able to be very aggressive in advancing the field, and defining the next generation of diagnostic tools and treatments for autism, both behavioral and pharmacological,” says Dr. Buxbaum.

THE EIGHTEENTH ANNUAL

Advances in Autism Conference

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