



Novel Consortium Meets

Working to Sequence DNA from 10,000 People with Autism

Organized by Dr. Joseph Buxbaum, Director of the Seaver Autism Center, and Dr. Matthew State, Deputy Chairman for Research in the Department of Psychiatry at Yale University School of Medicine, the Autism Sequencing Consortium (ASC) is composed of an international group of researchers who initially came together in 2010. Their goal is to facili-

tate the collection and analysis of whole exome sequence data (the segments of DNA that code for specific proteins) from as many as 10,000 individuals with autism, with the eventual aim of contributing to improved treatments.

The formation of this groundbreaking consortium is notable because the researchers agreed to

Continued on next page

IN THIS ISSUE

1 NOVEL CONSORTIUM SEAVER ON THE WEB

2 NOVEL CONSORTIUM (CONTINUED) EUSTACIA CUTLER DVD RECORDING

3 MEET THE FACULTY

4 SCIENCE AND SANDWICHES HOLIDAY PARTY



Founding members of the Autism Sequencing Consortium.

Seaver on the Web



The Seaver Autism Center is now on Facebook! Visit www.facebook.com/SeaverAutismCenter and “like” our page to learn about the latest research findings and autism-related events being hosted by the Seaver Autism Center.

Ways to Give

The Seaver Autism Center can now accept donations online! If you are interested, please visit <http://www.SeaverAutismCenter.org> and click on the “Ways to Give” button. We greatly appreciate the continued generosity that allows the Seaver Autism Center to make discoveries in autism genetics and clinical treatment.

Novel Consortium *Continued from previous page*

share data before it even existed – everyone involved in the ASC recognized that the data would be much more useful combined than if kept in individual institutions. To date, 21 groups have joined the ASC.

The initial stages of the formation of the group in February 2010 got off to an interesting start courtesy of Mother Nature, as Manhattan was snowed in and researchers had to cancel their trip. Not ones to give up, the group held a web conference instead. Participants included 18 scientists, as well as representatives from the National Institute of Mental Health (NIMH), Autism Speaks, the Simons Foundation, the Seaver Foundation, and the Hilibrand Foundation. Subsequently, in April 2010, the first face-to-face ASC meeting was held at the New York Academy of Medicine, and was generously supported by the Seaver Foundation.

This meeting was a great success, as it allowed the researchers and other representatives to conduct an in-depth ex-

amination of the initial objective for the ASC, which is to jointly analyze whole genome/whole exome data of autism samples and controls. Later in 2010, the ASC presented their goals and structure to Dr. Alan Guttmacher, the then newly appointed director of the National Institute for Child Health and Development.

As a next step, together with the ASC, Dr. Thomas Lehner, Director of the Office of Genomics Research Coordination and Chief of the Genomics Research Branch at the NIMH, organized the second face-to-face meeting at their Bethesda campus. At the meeting, the ASC presented an update on their initial goals, as well as new cutting-edge research. The attendees discussed these presentations and focused on where the field should be going in the short term, and in five years from now.

Presentations were given by Drs. Thomas Lehner, Joseph Buxbaum, Thomas Insel (NIMH), Mark Daly (Broad Institute), Steve Scherer (The Hospital for Sick Children, Toronto)

Evan Eichler (University of Washington), Bernie Devlin (University of Pittsburgh), Christina Hultman (Karolinska Institute) and Eric Schadt (Mount Sinai School of Medicine). Breakout groups focused on the technology used to sequence and analyze DNA samples, obtaining samples, and what the next challenges are. Dr. State presented a meeting summary at the end of the day.

As discussed at the meeting, the long-term goal of the ASC, “to identify at least 50% of the genetic risk for autism and translate this into improved patient care,” is viewed as reasonable and obtainable. The ASC is continuing their dialog on an ongoing basis, and individual sites are in direct contact to analyze emerging whole exome sequencing data in order to carry the group’s mission forward. ■



To view a videocast of the meeting at the NIH, please visit <http://videocast.nih.gov/summary.asp?Live=10546>.



Eustacia Cutler, author of *Thorn in My Pocket: Temple Grandin’s Mother Tells the Family Story*, with Dr. Alex Kolevzon at a recording of her new DVD. Ms. Cutler and her daughter Temple have recently established the Temple Grandin/Eustacia Cutler Autism Fund. Recognizing that autism is a neurological disorder that creates social disorder at home and in the community, their fund will be seeking donations to support and guide all family members and increase community understanding.

Information about how to obtain a copy of the DVD will be in a future issue of this newsletter and on www.SeaverAutismCenter.org when it becomes available.



Meet the Faculty

David Grodberg, M.D. is a child and adolescent psychiatrist and an Assistant Professor in the Department of Psychiatry at Mount Sinai School of Medicine. Dr. Grodberg's development from clinician to autism researcher is a result of extensive experience with autistic patients during his clinical training and as associate medical director at the Association for Metroarea Autistic Children, an Applied Behavior Analysis school in New York City. Despite significant advances in genetics, neuroimaging, and clinical trials, Dr. Grodberg realized the tremendous need for continued research into the neurobiology of the disorder. He also became acutely aware of the need for rapid and reliable standardized diagnostic assessment in non-research settings. These two areas of need shaped Dr. Grodberg's research endeavors.

Dr. Grodberg's interests in the etiology of autistic patients' sensory symptoms and stereotypic motor mannerisms led him to develop the hypotheses for his functional MRI study. He questioned whether this group of symptoms reflects functional abnormalities in cerebellar and somatosensory brain structures. Identification of a neurofunctional biomarker could lead to better identification of a subgroup of patients that may be studied further.

To pursue this hypothesis, Dr. Grodberg was awarded a T32 National Research Service Award fellowship at the Seaver Autism Center from July 2007 until June 2009. The T32 also gave him the opportunity to launch the functional MRI protocol, which holds promise to shed light

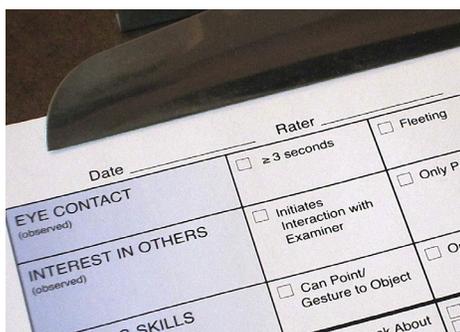
on the neurobiology of sensory symptoms in autistic patients.

Since joining the Seaver Autism Center as a full-time assistant professor in July 2009, Dr. Grodberg initiated another research project, which involves the creation and development of the Autism Mental Status Exam (AMSE). He realized there was a vital need for an autism-focused mental status exam for use in both research and non-research settings that was shorter and easier to perform than the more comprehensive tests that can take many hours to administer. In the fall of 2009, he began to develop the AMSE, which is an eight-item



observational assessment that takes place in the context of the clinical examination and produces a total score of 0-16.

In the winter of 2010, the AMSE was incorporated into the core assessment protocol at the Seaver Autism Center. Dr. Grodberg, along with Paige Weinger, a Ph.D. candidate, Latha Soorya, Ph.D., Alexander Kolevzon, M.D. and the rest of the Seaver team, promptly initiated a research protocol to analyze the reliability, validity, sensitivity and specificity of the AMSE to determine if specific cutoff scores predict how a subject will be classified on the gold standard observational assessment, the Autism Diagnostic Observation Schedule (ADOS). A recent publication in the *Journal of Autism and Developmental Disabilities* reports that the AMSE has excellent classification accuracy when compared to the ADOS in a high-risk population. The AMSE has been presented at major national and international conferences, including the American Academy of Child and Adolescent Psychiatry and the European Psychiatric Association. The AMSE will soon be available through the Seaver Autism Center web site. ■



Question sample from the Autism Mental Status Exam.



seaver autism center
for Research & Treatment at Mount Sinai

One Gustave L. Levy Place
Box 1668
New York, NY 10029



The Autism Science Foundation will be hosting Science & Sandwiches with the Seaver Autism Center on Friday, January 27, 2012. Families and individuals are invited to hear autism-related information from leading researchers. Please send an email to jessica.brownfeld@mssm.edu if you would like to attend.



The Fifth Annual Connections of New York-Seaver Autism Center Holiday Party was hosted at Mount Sinai by donor Rob Greenfeld and his team. Families, Seaver faculty and staff, as well as members of Mr. Greenfeld's company attended the holiday celebration. **JP Laramée** (left) entertained party guests with a magic show. Thank you very much to Mr. Greenfeld and his team for their continued support of the Seaver Autism Center!

• **THE SEAVER AUTISM CENTER NEWSLETTER** brings you timely updates about new developments related to research and treatment of autism spectrum disorders, as well as activities at the Seaver Autism Center. To be placed on our mailing list, please contact SeaverCenterEditor@mssm.edu or The Seaver Autism Center, Mount Sinai School of Medicine, One Gustave L. Levy Place, Box 1668, New York, NY 10029. Our phone number is 212.241.0961 and our web site is www.SeaverAutismCenter.org.

• **GOING GREEN:** Please send your email address to SeaverCenterEditor@mssm.edu to receive future issues of the Seaver Center Newsletter. If you don't have an email address, you will continue to receive the newsletter in the mail.



PARTIALLY
RECYCLED PAPER