Understanding the Roots of Autism

Specific cellular pathways, along which genetic mutations occur, appear to play a key role in the development of autism spectrum disorders (ASD), according to new research from Icahn School of Medicine at Mount Sinai.

The findings, published online in the April 24, 2014, issue of *The American Journal of Human Genetics*, provide scientists with a better understanding of the complex genetic architecture involved in ASD, which has its roots in early brain development.

The study was led by Dalila Pinto, PhD, Assistant Professor of Psychiatry, and Genetics and Genomic Sciences at Icahn School of Medicine, as part of the Autism Genome Project Consortium, and examined 2,446 individuals with ASD and their parents in the United States, Canada, and Europe. The team isolated DNA from the participants’ blood samples or cell lines genotyped with high-resolution microarrays, and examined the copy-number variation (CNV)—or deletions or duplications in sections of DNA—in the samples.

A New Master’s Program in Health Care Delivery Leadership

The Icahn School of Medicine at Mount Sinai is launching a unique Master’s program specifically geared to give health care leaders new knowledge and skills as they confront the challenges of delivering patient care in an era of unprecedented reform. The Master’s Program in Health Care Delivery Leadership is offered through the Department of Health Evidence and Policy and is currently accepting applications for its inaugural cohort that begins this fall.

Recruitment will occur among the ranks of senior leaders and managers within the Mount Sinai Health System, as well as other national, and international, health care institutions. The program requires successful applicants to possess at least seven years of managerial-level health care experience. It is built upon a core belief that effective solutions to current challenges in health care delivery require...
A New Master’s Program in Health Care Delivery Leadership

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Students enrolled in the program will have access to the expertise and resources of a top 20 school of medicine that resides within a comprehensive urban health system serving a highly diverse patient population. “We see this program as a logical outgrowth of the innovative initiatives in place across the Mount Sinai Health System, and as a reinforcement of Mount Sinai’s role at the center of academic research and education during this transformational time,” says Dennis S. Charney, MD, Anne and Joel Ehrenkranz Dean, Icahn School of Medicine at Mount Sinai and President for Academic Affairs, Mount Sinai Health System.

Students will have direct access to national leaders in health care reform, gain practical experience, and benefit from the strength of the faculty in the Department of Health Evidence and Policy, throughout the School of Medicine and the Graduate School of Biomedical Sciences. To facilitate convenience, the majority of the rigorous coursework will be delivered online.

Within a week of announcing the program in late April, the School of Medicine received more than 150 inquiries and multiple applications. Applicants will receive a comprehensive evaluation for admission based upon their academic records, professional experiences, leadership capabilities, major accomplishments, and motivation to complete the demanding program.

The program is co-led by Alan J. Moskowitz, MD, Professor and Vice Chair, Health Evidence and Policy, and Professor of Medicine; and Brian J. Nickerson, PhD. For additional information, visit www.icahn.mssm.edu/mshcdl.

BioMe Biobank Celebrates 30,000 Participants

More than 300 employees and patients of the Mount Sinai Health System recently gathered in the Guggenheim Atrium to celebrate the 30,000th participant in the BioMe Biobank. The Biobank collects de-identified DNA and plasma used for a variety of research purposes from consenting patients.

The celebration took place on Thursday, April 24, National DNA Day, as participants gathered around a giant cake in the shape of a DNA double helix that was covered in a sweet gray fondant. One side of the cake was chocolate and raspberry and the other side was vanilla and strawberry. Two-hundred-fifty test tubes filled with cranberry juice were made to resemble the tubes of blood that are collected from BioMe participants.

When patients agree to participate in the Biobank program, their extracted genetic material is linked to their electronic medical records. Their DNA and plasma is frozen and banked until it is selected for a variety of patient-centered, institutional review board-approved research projects, with the ultimate goal of creating personalized treatments.
Understanding the Roots of Autism

The team’s results confirmed previous findings that people with autism have an excess of rare genetic deletions and duplications affecting genes, when compared with people who do not have the disorder. In addition, the investigators reported that some of these genetic changes were inherited, while others were de novo and not observed in the parents.

ASD is estimated to occur in 1 out of every 68 births, according to the U.S. Centers for Disease Control and Prevention. It is a host of complex disorders characterized by difficulties in communication and social interaction, and repetitive behaviors.

Dr. Pinto says her team began to analyze the pathways in which genetic activity occurs after asking the following question: “If different genes can be altered in different individuals with ASD, why does it manifest itself so similarly in individuals?”

She says, “We found that changes can affect different genes but always the same interconnected pathways. Some nodes of the pathway or network are more important than others. But when the hubs are dysregulated, the system gets disturbed, and the result will determine the clinical phenotype of the child.”

In a number of individuals with ASD, the researchers discovered that their respective CNVs involved a gene that indicated greater risk for developing epilepsy, demonstrating that identifying genetic alterations for ASD may indicate other medical conditions.

The recent findings expanded upon an earlier study led by Dr. Pinto in 2010. The next step, she says, will be an even larger study that enables her team to find more genes and additional sections of the pathways.

Her lab is searching for genetic overlaps between ASD, epilepsies, and intellectual disabilities. Understanding the biology behind autism, she adds, will help scientists improve early diagnosis and intervention.
**Events**

### The Douglas West Endowed Memorial Visiting Lecture

Author Katy Butler presents “Knocking on Heaven’s Door,” based upon her experience shepherding her parents through their final declines. For more information, contact Katie Madden at 212-241-0117.

Sponsored by the Lilian and Benjamin Hertzberg Palliative Care Institute of the Brookdale Department of Geriatrics and Palliative Medicine.

**Monday, May 19**

5:30 pm

The Mount Sinai Hospital

Goldwurm Auditorium

### Grand Rounds Neurology

Mark Hallett, MD, Chief, Human Motor Control Section, National Institutes of Health, presents “Pathophysiology of Dystonia.”

**Wednesday, May 14**

8 – 9 am

The Mount Sinai Hospital

Hess Center

Seminar Room B

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### Grand Rounds Diabetes

Charles Mobbs, PhD, Professor, Neuroscience, Medicine, and Geriatrics and Palliative Medicine, presents “Hypoglycemia in Diabetes: Novel Mechanisms and Therapies.”

**Thursday, May 15**

8:30 – 9:30 am

The Mount Sinai Hospital

Atran Building AB4-11

### Grand Rounds Medicine

Philip Mackowiak, MD, Professor and Vice Chair, Department of Medicine, University of Maryland School of Medicine, presents “Mozart’s Fatal Anasarca.”

**Tuesday, May 20**

8:30 am

Mount Sinai Beth Israel

Podell Auditorium

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### Free Melanoma Cancer Screening

Staff and the public are invited as walk-ins to receive a free total body skin examination. To learn more, call 212-523-3888.

**Mount Sinai Beth Israel**

Saturday, May 31

10 am – 2 pm

Phillips Ambulatory Care Center

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### Announcements

#### Nurses Week 2014

Joyce Fitzpatrick, PhD, RN, FAAN, Adjunct Professor, Icahn School of Medicine at Mount Sinai, Professor of Nursing, Case Western Reserve University, will present the keynote address.

**The Mount Sinai Hospital**

Monday, May 12

Presentation: Noon

Hatch Auditorium

Taped Keynote: 10 pm

Guggenheim Pavilion 2A

#### Computer-Based Medical Systems Symposium

This year’s 27th IEEE International Symposium on Computer-Based Medical Systems (CBMS 2014) will be held at Icahn School of Medicine at Mount Sinai. Attracting a worldwide audience, CBMS 2014 is one of the premier conferences for computing in health care.

Email Marina Krol, PhD, at marina.krol@mountsinai.org to register. To learn more, visit www.sites.ieee.org/cbms.

**Tuesday, May 27 – Thursday, May 29**

The Mount Sinai Hospital Campus

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### Calling all Mount Sinai Runners

The Mount Sinai Health System’s running group is gearing up for its June kickoff, when members begin to meet weekly in Central Park on Tuesday evenings until November. Training will include pacing and handling hills as runners work their way up to 20 miles.

“The group is open to runners from every department and every hospital within the Mount Sinai Health System,” says the group’s coordinator, David Kaplan, MPA, Vice Chairman, Administration and Finance, Health System Administrator, Surgical Services, and Academic Instructor of Surgery.

Approximately 50 Mount Sinai runners are expected to participate in the Brooklyn Half marathon on Saturday, May 17, stretching from Prospect Park to the Coney Island boardwalk.

To learn more, call David Kaplan at 212-241-1941, or email david.kaplan@mountsinai.org.

*Photo: In March, Susanne Erni, Digital Projects and Analytics Manager, and other members of the Mount Sinai running group participated in the New York Half marathon through Manhattan.*