



**Mount
Sinai**

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Mount Sinai Introduces New Genetic Screening Tests



The Mount Sinai Genetic Testing Laboratory's Executive Director, Lisa Edelmann, PhD, left, and Director, Ruth Kornreich, PhD

The Mount Sinai Genetic Testing Laboratory in January introduced a new panel of comprehensive pan-ethnic carrier screening tests for 281 genetic disorders, the largest currently available. Mount Sinai's NextStep Carrier Screening also includes the most comprehensive panel of tests for 96 diseases found in the Ashkenazi Jewish population and is the first of its kind to address the largely overlooked needs of the Sephardi and Mizrahi Jewish populations.

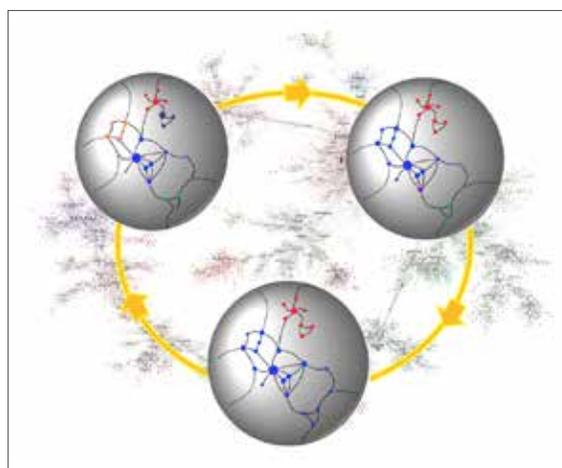
"Building on years of in-house genetic research and technology adaptation in our clinical laboratory, we created tests that not only expand the number of diseases screened, but also increase the breadth of coverage, to improve carrier detection rates and provide more accurate residual risk estimates to patients," says Lisa Edelmann, PhD, Executive Director of the Mount Sinai Genetic Testing Laboratory within the Department of Genetics and Genomic Sciences.

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Big Data Tools Help Decipher Disease Progression

Two new Big Data analysis tools that help pinpoint specific genes that are actively involved in disease progression were recently made available to the public by scientists in the Multiscale Network Modeling Laboratory at the Icahn School of Medicine at Mount Sinai.

The team, led by Bin Zhang, PhD, Associate Professor in the Department of Genetics and Genomic Sciences, published the pair of algorithm-based tools online in November 2015 in *PLoS Computational Biology* and in *Scientific Reports*, a *Nature* publication. The open-source tools



The MEGENA tool has 3D spheres that help uncover precise network clusters associated with disease progression.

are available to all researchers who wish to gain a better understanding of disease mechanisms in order to develop more effective drugs and create individualized treatments.

One of the tools, MEGENA (Multiscale Embedded Gene Co-Expression Network Analysis), enables researchers to project massive amounts of gene expression data from complex diseases such as cancer onto a three-dimensional sphere to uncover highly precise network clusters associated with disease progression.

"In reality, the boundaries of clusters formulated by traditional approaches

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The new carrier tests are designed to help prospective parents more accurately identify whether they carry certain genetic mutations that might affect their offspring. Jewish populations, particularly Ashkenazi Jews, are at increased risk for a wide range of genetic disorders, including Gaucher disease, Tay-Sachs disease, Niemann-Pick disease, Bloom syndrome, and Factor XI deficiency.

According to Dr. Edelman, between 50 percent and 70 percent of Jewish individuals will screen positive for at least one of the conditions in the new Comprehensive Jewish Panel. “Most of these conditions involve childhood onset and are severely debilitating,” Dr. Edelman says.

In order to develop additional tests and provide patients with more accurate residual risk estimates, Mount Sinai’s Genetic Testing Lab worked closely with clinical geneticists who see patients for rare diseases and with the bioinformatics team at the Icahn Institute for Genomics and Multiscale Biology.

“We leveraged our expertise with next-generation DNA sequencing along with several other state-of-the-art technologies to deliver the

best available carrier screening tests to our physicians and their patients,” says Eric Schadt, PhD, the Jean C. and James W. Crystal Professor of Genomics at the Icahn School of Medicine at Mount Sinai, and Founding Director of the Icahn Institute for Genomics and Multiscale Biology.

Six testing technologies are used in parallel to ensure the highest detection rate for each gene. The lab is able to examine a person’s DNA or RNA and the structure of his or her chromosomes, in addition to evaluating, diagnosing, and monitoring treatment of patients with inherited metabolic diseases.

Over the past two decades, the Mount Sinai Genetic Testing Laboratory has become a market leader in the field of carrier testing for genetic diseases.

The lab processes tens of thousands of samples each year. Geneticists and physicians who specialize in obstetrics, maternal fetal medicine, in vitro fertilization, and pediatrics use Mount Sinai’s genetic tests to help them make critical care decisions. Clients also include other major health systems in the New York metropolitan area.

Genetic Testing Under Way in Connecticut

In its first full year of operation, the Mount Sinai Genetic Testing Laboratory in Branford, Connecticut, has become an integral part of the Mount Sinai Health System’s efforts to better diagnose and treat disease.

The 16,400-square-foot facility, located 85 miles from New York City, has the high-throughput equipment to sequence thousands of samples monthly to uncover variations in DNA that code for Alzheimer’s and coronary disease, and cancer, among other diseases.

“We’ve demonstrated that we’re on the cutting edge of today’s data-driven medicine,” says Todd Arnold, PhD, Managing Director of the Genetic Testing Laboratory in Branford. “Through our ability to generate data from hundreds of samples a day, we’re giving researchers and clinicians a fast and effective way to identify variants that can be linked to disease.”

A milestone for the laboratory was its certification in November from the Centers for Medicare and Medicaid Services and the State of Connecticut to begin processing clinical samples originating from many states. Licensing is expected to be secured for New York State later this year. As part of that effort, all oncologic sequencing will be transferred to Branford from the Mount Sinai Genetic Testing Laboratory in Manhattan.

The Branford facility is gearing up to handle what ultimately



In Mount Sinai’s Branford, Connecticut, laboratory, Research Associate Courtney Pietropaolo prepares DNA samples for sequencing.

could be several hundred thousand samples as part of the Resilience Project. This vast Mount Sinai research effort will ask for donations of DNA in order to find individuals who have managed to stay healthy despite rare genetic mutations that, according to the medical textbooks, should have predisposed them to catastrophic illness.

A Milestone for the Phillips Ambulatory Care Center

Mount Sinai Beth Israel employees joined together on Friday, January 22, for a groundbreaking ceremony kicking off renovations to the Phillips Ambulatory Care Center's (PACC) lobby and façade. This marks the first step in an overall renovation and redesign of the facility.

"Mount Sinai continues to invest in the future state of PACC as a first-class ambulatory care center in the heart of downtown Manhattan," says Burton Drayer, MD, Chief Executive Officer, Mount Sinai Doctors Faculty Practice, and Dean for Clinical Affairs, Icahn School of Medicine at Mount Sinai. "Ultimately, we expect that a modernized facility will reflect the high level of quality care our talented staff provides our patients."

The ceremony included remarks by hospital leadership highlighting the structural and cosmetic changes that PACC is undergoing and provided attendees the opportunity to ask questions. A presentation of design renderings displayed plans for a modern PACC lobby that will feature a contemporary staircase, which will offer patients and staff direct access to the second floor, as well as other efficiencies. Already, the Duane Reade pharmacy has been relocated from the lobby to the second floor; the elevators are undergoing improvements; and construction has begun on the building's façade.



From left: Paul Zucker, Vice President, Ambulatory Operations, Phillips Ambulatory Care Center (PACC); Kelly Cassano, DO, Chief of Ambulatory Care, Mount Sinai Beth Israel, Associate Dean, Clinical Affairs, Icahn School of Medicine at Mount Sinai; Burton Drayer, MD; Susan Somerville, RN, President, Mount Sinai Beth Israel; Stacy Coleman, Vice President of Operations, PACC; and Elizabeth Sellman, MPA, Chief Operating Officer, Mount Sinai Beth Israel.

PACC has been providing comprehensive outpatient services to the New York community since 1996. Its services include primary and specialty care, onsite diagnostic services, and a state-of-the-art ambulatory surgery center.

› Big Data Tools Help Decipher Disease Progression *(continued from page 1)*

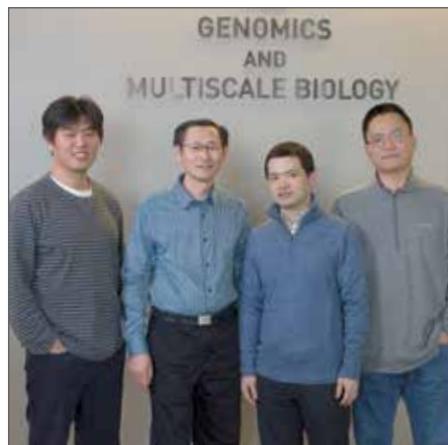
aren't so clear, and many genes may be excluded from a cluster at a certain threshold of compactness when they shouldn't," says Dr. Zhang. "MEGENA resolves the problem by detecting clusters at multiple compactness multiscales that are naturally determined by the data. When you remove redundant correlations, you get a more reliable, more robust network structure."

The other software tool, SuperExactTest, helps scientists determine the significance of similarities among multiple data sets. SuperExactTest allows researchers to compare three or more data sets, a feat that is new in the field of statistics. The tool has wide application and is expected to yield valuable new insights.

"Before we came up with SuperExactTest, nobody could exactly calculate the statistical significance of intersections among multiple sets of genes," according to Dr. Zhang.

Mount Sinai researchers have already

used MEGENA to uncover novel regulatory targets in breast and lung cancers after testing a large amount of genomic data of U.S. patients from The Cancer Genome Atlas (TCGA), a master data repository assembled by the National Institutes of Health. MEGENA showed improved performance over well-established clustering methods



Big Data developers, from left: Won-min Song, PhD, Senior Scientist; Bin Zhang, PhD; Minghui Wang, PhD, Postdoctoral Fellow; and Yongzhong Zhao, PhD, Senior Scientist

and gene co-expression network construction approaches.

SuperExactTest led to the discovery that a set of nine genes is the most critical in determining whether a person is predisposed to getting cancer. The pioneering software allowed Mount Sinai's researchers to determine the significance of this core set of genes that are common to seven mutated gene sets in cancer.

"Our team is dedicated to crafting high-performance analysis tools and to sharing those resources with the broader genomics community to help us all generate the best possible results," says Eric Schadt, PhD, the Jean C. and James W. Crystal Professor of Genomics at the Icahn School of Medicine at Mount Sinai, and Founding Director of the Icahn Institute for Genomics and Multiscale Biology. "These new tools demonstrate thoughtful and creative solutions to computational challenges faced by scientists around the world, and I look forward to seeing what the community will accomplish with them."

Around the Health System

Excellence in Nursing Leadership

Two clinical nurse managers at The Mount Sinai Hospital recently received the Association of the Attending Staff (AAS) 2015 Award for Excellence in Nursing Leadership. The recipients—Rita Jakubowski, MSN, RN, Clinical Program Manager, Bone Marrow Transplant; and Rosario Marasigan, BSN, RN, Clinical Nurse Manager, Cardiac Catheterization Laboratory—were honored as role models who enhance the quality of the practice environment. They were recognized for their compassion, motivation, clinical expertise, and leadership, all in an effort to provide exemplary patient care. The awards were presented by AAS President Eric Neibart, MD, Assistant Clinical Professor of Medicine (Infectious Diseases), Icahn School of Medicine at Mount Sinai.



From left: Rita Jakubowski, MSN, RN; Carol Porter, DNP, RN, FAAN, Senior Vice President and Chief Nursing Officer, The Mount Sinai Hospital, Edgar M. Cullman, Sr. Chair of the Department of Nursing, Associate Dean of Nursing Research and Education, Icahn School of Medicine at Mount Sinai; and Rosario Marasigan, BSN, RN.



Brad Beckstrom, second from left, and Daniel Reyes, Deputy Executive Director, New York Common Pantry, far right, distribute food to Ramona Perez, left, and Jose Martell at the Bonifacio Cora Texidor Senior Center.

Working Together to Fight Hunger

The New York Common Pantry (NYCP), which has the long-standing support of the Mount Sinai Health System, recently began delivering nutritionally balanced food directly to community providers that serve or house senior citizens. Ultimately, about 13,500 seniors a month will receive food through this new program. NYCP already provides hot meals and support services to more than 49,000 individuals a year at the organization's 8 East 109th Street site. Brad Beckstrom, Senior Director of Government and Community Affairs, Mount Sinai Health System, who serves on the NYCP Board of Directors, says, "We strongly believe in the mission of the New York Common Pantry and provide financial support, give turkeys and hams at Thanksgiving and Christmas, partner with them on promoting healthy eating, and distribute coupons for shopping at the green markets. Many Mount Sinai volunteers also offer their services."

Observing World AIDS Day

In honor of World AIDS Day on Tuesday, December 1, the Mount Sinai Institute for Advanced Medicine teamed with the Gay Men's Health Crisis to present "World AIDS Day: Focus on the Transgender Community" at Mount Sinai West. The event featured speaker Octavia Y. Lewis, MPA, Patient Services Manager, Community Healthcare Network, and a performance by Tony Award winner Michael Cerveris and the musical group Loose Cattle. Among other activities: information booths and HIV testing were available at Mount Sinai West and at the Peter Krueger Clinic at Mount Sinai Beth Israel; The Mount Sinai Hospital distributed literature on Pre-Exposure Prophylaxis (PrEP), Post-Exposure Prophylaxis (PEP), and other HIV-related material; and the Mount Sinai Comprehensive Health Program-Downtown conducted additional HIV testing.



Staff from the Mount Sinai Institute for Advanced Medicine's Peter Krueger Clinic Care Coordination Program distributed giveaways and HIV information and provided HIV testing at Mount Sinai Beth Israel.

Helping to Establish Sustainable Health Care in Liberia

After suspending travel to Liberia during the largest outbreak of Ebola in history, faculty at the Icahn School of Medicine at Mount Sinai resumed their teaching trips to the West African country last fall, with renewed efforts to improve women's health.

Led by Ann Marie Beddoe, MD, Assistant Professor, Obstetrics, Gynecology and Reproductive Science, members of the Mount Sinai community have undertaken several initiatives in Liberia since they began working there in 2008. They are helping to train the country's first residents in obstetrics and gynecology and have applied for a grant from the National Institutes of Health to help build a cancer center. They have also trained nurses to conduct human papillomavirus (HPV) screenings and counsel patients.

The Icahn School of Medicine and several U.S. universities, including Harvard University and Massachusetts Institute of Technology, belong to a consortium that is helping Liberia establish educational programs in basic science, radiation therapy, and pathology, as well.

The overriding goal of all of these programs is to help the country build its own

sustainable, health care infrastructure, which was destroyed in two successive civil wars, the last of which ended in 2003. Following the Ebola outbreak in 2014, Mount Sinai faculty members no longer have direct contact with Liberian patients. Their trips are entirely educational.

Dr. Beddoe's first visit to Liberia in 2008 was organized by Jeffrey S. Freed, MD, an Associate Clinical Professor in the Department of Surgery. At the time, she says there were 22 doctors serving the entire nation of 4 million people. Only one of those doctors specialized in OBGYN care. She says Mount Sinai's involvement and the support of Michael Brodman, MD, the Ellen and Howard C. Katz Chairman's Chair in the Department of Obstetrics, Gynecology and Reproductive Science, are helping to pave the way for Liberia's first class of OBGYN residents to graduate in 2017.

Unlike other impoverished countries, Liberia had no health care infrastructure in 2008. "War had decimated everything," says Dr. Beddoe, who is also Director, Chemotherapy Infusion Service, Icahn School of Medicine at Mount Sinai. One CT scan machine served the entire country and there were no ventilators. Patients who were wealthy



Ann Marie Beddoe, MD, right, and Eileen Solomon in Liberia

and healthy enough traveled to nearby Ghana to receive radiation treatment. With the assistance of Eileen Solomon, Director, Special Events at the Mount Sinai Health System, who, as a volunteer, has helped facilitate 15 trips to the country, Mount Sinai has been able to make inroads.

"I saw so many women with cervical cancer who were being treated for infections. The doctors were unaware of cervical cancer," says Dr. Beddoe. She and Peter R. Dottino, MD, Director of the Division of Gynecologic Oncology at the Mount Sinai Health System, teamed with a local doctor to initiate the country's first chemotherapy program at the John F. Kennedy Medical Center in the capital, Monrovia, where patients were treated for free. Liberia's Ministry of Health is planning to open a dedicated cancer hospital this year.

In an upcoming initiative, Dr. Beddoe plans to train 40 health care workers to serve as HPV specialists who will build awareness about the importance of cervical cancer screenings and early detection, when the cancer can be treated successfully. To assist in this effort, the biomedical company Qiagen has donated 2,000 free HPV screening kits and a machine that reads same-day test results. Dr. Beddoe says Mount Sinai has applied for 8,000 more tests.

"If we can get the Ministry of Health to do screenings on an ongoing basis, we know we can save women's lives," she says.



At the John F. Kennedy Medical Center in Monrovia, Liberia, health officials, doctors, residents, and medical students gathered for grand rounds on the importance of research that were presented by Mount Sinai's OBGYN team.

Mount Sinai Health System Hosting Heart-Health Activities

Staff and the public are invited to participate in a number of activities that promote heart health, an annual initiative in February that seeks to educate individuals about the risk of cardiovascular disease.

Women's Heart NY

The following events are sponsored by Women's Heart NY, a Mount Sinai Health System comprehensive heart program. Registration is required. To register, call 212-523-9663 or email strignano@chpnet.org.

Support Group Meetings

Facilitated by Sydney Fischer, MSW, these meetings are for Women's Heart NY patients seeking support in navigating challenges related to heart-healthy living and/or recovering from cardiovascular surgery.

Mondays, February 8 and 22

6 – 7:30 pm

**Mount Sinai Beth Israel
Phillips Ambulatory Care Center
Bogart Conference Room**

"Heart Smart" Education Course

This free, six-session heart-health program is sponsored in partnership with New York City Parks and Recreation. Sessions will include lectures presented by Mount Sinai Heart cardiologists and dietitians, as well as exercise professionals, including yoga instructors. *This program is made possible, in part, by the TD Bank Charitable Foundation.*

Wednesdays, February 10 - March 16

5 – 6:30 pm

**NYC Parks and Recreation
Recreation Center 54
348 East 54th Street**

Session 1 – February 10: Heart Health Information

(Also, stop by between 1 – 4 pm for free cholesterol and glucose screenings sponsored by the Mount Sinai Department of Government and Community Affairs and Karpas Health Information Center.)

Session 2 – February 17: Nutrition Part I

Session 3 – February 24: Exercise Part I

Session 4 – March 2: Stress Recognition and Management

Session 5 – March 9: Nutrition Part II

Session 6 – March 16: Exercise Part II and Wrap-up

Exercise Planning for Heart Health

Saturday, February 13

Noon – 1:30 pm

**Asser Levy Recreation Center
392 Asser Levy Place at East 23rd Street
and FDR Drive**

Community Blood Pressure Screening Program

Free blood pressure monitoring and consultation with a medical professional. *Sponsored by Mount Sinai Heart*

Wednesday, February 10

10:30 – 11:30 am

Hamilton Senior Center

141 West 73rd Street between Amsterdam and Columbus Avenues

For more information, call 212-787-7710.



**Mount
Sinai
Heart**

Eat Heartily with Chef Rocco Sacramone

Mount Sinai Queens, the United Community Civic Association, and Central Sushi Bar and Lounge present a heart-healthy cooking demonstration and tasting by Zagat-rated Trattoria L'incontro's chef-owner Rocco Sacramone, who will demonstrate ways to preserve taste when preparing heart-healthy meals.

RSVP is required:

Visit [www.mshq.org/
community/events](http://www.mshq.org/community/events) or
call 800-YOUR-MDS
(800-968-7637).

Thursday, February 25

6:30 pm

**Central Sushi Bar
and Lounge
20-30 Steinway Street
Astoria, New York**



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