Consortium Sheds New Light on Brain Disorders

More than two dozen researchers at the Icahn School of Medicine at Mount Sinai are advancing brain science by mapping the complex molecular underpinnings of autism spectrum disorder, schizophrenia, and bipolar disorder through their work in the National Institute of Mental Health’s (NIMH) PsychENCODE Consortium. Since this work began in 2015, their contributions—and that of their PsychENCODE colleagues from 14 other U.S. institutions—have helped identify several hundred new risk genes for mental disorders. The research has also revealed critical time windows during brain development when these genes can influence the disease process.

In December, the Consortium published its initial findings in 10 studies that appeared in Science, Science Translational Medicine, and Science Advances. The researchers analyzed more than 2,000 postmortem brain samples from people with no psychiatric conditions and those with schizophrenia, autism, and bipolar disorder. They created and then integrated data sets that included information on DNA variations and gene expression for about 32,000 cells from major regions of the brain. Then the investigators employed machine learning to create a predictive model of risk for the psychiatric disorders. Their seminal findings received an enthusiastic response from the NIMH. “The PsychENCODE project came through,” said Thomas Lehner, PhD, MPH, Director of the Office of Genomic Research Coordination at the NIMH. “We’re at the beginning—I cannot

Dubin Gala Honors Champions of Breast Cancer Care and Awareness

A leading physician-scientist and three “passionate champions” of breast cancer awareness were honored by the Dubin Breast Center of The Tisch Cancer Institute at the Mount Sinai Health System at its eighth annual benefit. The celebratory event, held on Monday, December 10, at the Ziegfeld Ballroom in Manhattan, attracted 520 guests and raised $2.6 million to support the Center’s breast health and treatment programs.

The three “champions” were Melissa Spohler, Meredith Shepherd, and their mother, Priscilla Alexander—all diagnosed with breast cancer in their 50s. “This is a family of women who have strength. They have integrity, they are kind, they give to others,” says Eva Andersson-Dubin, MD, founder of the Dubin Breast Center and Mount Sinai Health System Trustee, who presented awards to the honorees with Elisa Port, MD, FACS, the Center’s Director. “These are women we should all look up to as role models to see how they deal with breast cancer.”

Also honored at the gala was a “spectacular clinical investigator,” Amy Tiersten, MD, Clinical Director of Breast Medical Oncology, The Mount Sinai Hospital, and Professor

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overstate how early we are. But I can confidently say that for the first time we have a beginning of an understanding of the biology—the molecular pathophysiology of mental disorders—of schizophrenia, and bipolar and autism spectrum disorder.”

NIMH Program Director Geetha Senthil, PhD, added that the massive scope of the project required a “concerted effort. Many investigators had to come together and do this collectively.” While the mental disorders in the studies are distinct, Dr. Senthil said, “There are some aspects where the biology is similar. The genes interact with each other in a way to influence the disease process. If we can find biological clues early on, we can intervene early on. While we are building and generating more data, analyzing this data to find basic mechanisms, there’s an opportunity also for drug discovery.”

The 10 papers published by the PsychENCODE Consortium were dedicated to the late Pamela Sklar, MD, PhD, former Chair of the Department of Genetics and Genomic Sciences at the Icahn School of Medicine, and a pioneer in genomic brain research, who was an early leader of the NIMH effort. The Icahn School of Medicine last year renamed the division she created the Pamela Sklar Division of Psychiatric Genomics.

Mount Sinai laboratories within The Friedman Brain Institute, The Seaver Autism Center for Research and Treatment, the Department of Psychiatry, The Mindich Child Health and Development Institute, the Department of Genetics and Genomic Sciences, the Department of Neuroscience, and the Icahn Institute for Data Science and Genomic Technology were involved in the PsychENCODE Consortium.

“Mount Sinai serves as one of the lead sites in this national consortium. The discoveries that are being made by our scientists and their colleagues at other major institutions are moving us closer to understanding and finding treatments for these devastating brain disorders,” says Eric J. Nestler, MD, PhD, Nash Family Professor of Neuroscience, Director of The Friedman Brain Institute, and Dean for Academic and Scientific Affairs, Icahn School of Medicine at Mount Sinai.

The scientific team discovered that early development is associated with major changes in the spatial organization of DNA inside of brain cells. These changes in how the chromosomal material is packed seem to disproportionately affect DNA sequences linked to schizophrenia heritability risk and provide new insights into the genetic causes underlying this disease.

The study, which was conceived and executed at the Icahn School of Medicine, included senior authors Schahram Akbarian, MD, PhD, Professor, Psychiatry, and Neuroscience; and Kristen Brennand, PhD, Associate Professor, Neuroscience, Genetics and Genomic Sciences, and Psychiatry; Prashanth Rajarajan, MD/PhD candidate; and Schahram Akbarian, MD, PhD, Professor, Psychiatry, and Neuroscience.

“Eighteen years after fully sequencing the human genome, we still understand very little about how it actually comes to life. Our study, and the others that were published, are beginning to unravel more nuances than previously imagined...”

– Prashanth Rajarajan, MD/PhD candidate

Unraveling the Complexity of the Human Brain

“Exploring how the human genome is folded and packaged into the nucleus of each of our billions of brain cells was both awe-inspiring and humbling at the same time,” says Prashanth Rajarajan, MD/PhD candidate at the Icahn School of Medicine at Mount Sinai, who was first author on seminal brain research that was published in the December 14, 2018, issue of Science.

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First He Donated His Bone Marrow, Then He Gave Her a Kidney: How Two Strangers Became “Family Forever”

When Jeramy Davies was a senior at Texas Tech University in 2010, he helped organize a charity drive for Be the Match®, the national program that matches potential donors to those needing a bone marrow transplant. Little did he know that five years later he would donate his own bone marrow to a stranger in New Jersey—Kelly Ribeiro, who was being treated for lymphoma. And in 2018, he would give Ms. Ribeiro one of his kidneys, effectively saving her life twice.

Since hospital protocol forbids the exchange of any information between donor and recipient for one year, and then only if both parties agree, Mr. Davies and Ms. Ribeiro remained unknown to each other immediately following her successful bone marrow transplant. In 2016, Mr. Davies reached out to her and they began exchanging emails and text messages and speaking frequently by phone.

But over time, they both faced significant challenges. Ms. Ribeiro was dealing with kidney failure due to her previous condition. During a bout with pneumonia she fell into a coma. When she emerged, she began dialysis three times a week. “I never felt so sick and depleted as I did with kidney failure,” she says. “Every day, I could feel the life draining out of me.”

At the same time, Mr. Davies was helping his wife battle a brain tumor that would take her life in July 2017. “Kelly was great moral support for me,” Mr. Davies recalls. “She understood everything we were going through, and she was a huge source of strength.”

But Ms. Ribeiro did not want to burden him with her own struggle, and even as her kidney function dwindled to 19 percent, she did not ask for his help. Only after her mother told Mr. Davies how sick she was did he understand the full extent of Ms. Ribeiro’s situation. Immediately, he offered Ms. Ribeiro his kidney. Immediately, she turned him down.

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Dubin Gala Honors Champions of Breast Cancer Care and Awareness (continued from page 1)

of Medicine (Hematology and Medical Oncology), Icahn School of Medicine at Mount Sinai. “I have really poured a lot of lifeblood and energy into building a research program and clinical practice and collaborating with all the wonderful other physicians at the Dubin Breast Center,” says Dr. Tiersten, who specializes in metastatic breast cancer. “Through Dr. Tiersten’s research, we can provide our patients with the next generation of treatments—treatments that provide hope,” Dr. Port says.

The event included music by accomplished performers, including LaChanze, Ariana DeBose, and Storm Lever from Summer: The Donna Summer Musical on Broadway. And Ms. Alexander, 81, gave a moving speech about the Dubin Breast Center, saying that it did not yet exist when she was treated with cancer at age 53, but it now provides “great comfort” to her daughters and other patients.

“My daughters are part of a sisterhood at the Center,” Ms. Alexander says. “They have the good fortune of being watched over by the best professionals in their specialties, trained at the cutting edge of testing, diagnosis, treatment, emotional support, and ongoing research.” As a patient at another New York City hospital 28 years ago, Ms. Alexander had a double mastectomy and took an early test for a BRCA (breast cancer susceptibility) gene mutation. The test was negative. But because her mother and four of her six female cousins had all been treated for breast cancer, Ms. Alexander urged her four daughters to be vigilant. Ms. Shepherd says, “In hearing my family’s history, everyone’s first question is ‘Are you a BRCA family?’ I always say, ‘No, we’re a something family, it just hasn’t been found yet.’”

Ms. Spohler, 56, had at least one breast screening test every six months starting at age 40. In March 2016, a radiologist saw ambiguous “bright spots” on an MRI, and six months later an ultrasound identified breast cancer. “I was lucky because it was discovered when it was small,” Ms. Spohler says. “I had a double mastectomy because I didn’t want to live with the fear of it coming back.” Ms. Shepherd, 52, was diagnosed with breast cancer in August 2018. Her radiologist saw a small mass in her right breast, and a biopsy found that it was a benign growth called a papilloma. Ms. Shepherd says she went “right away” to Dr. Port, who had also treated her sister. As part of a preoperative check before removing the growth, Dr. Port ordered an MRI. That test discovered breast cancer unrelated to the papilloma, and Ms. Shepherd also had a double mastectomy.

All of the honorees say the key to early detection is awareness. “I would say to any woman that if you have breasts, you are potentially at risk,” Ms. Spohler says. “You have to monitor yourself. You have to know your own history and know your family’s history, because that is what might save you.”
Kidney: How Two Strangers Became “Family Forever”

“It didn’t feel right,” she says. But he persisted, and in late December 2018, she underwent a successful kidney transplant at The Mount Sinai Hospital. Since Ms. Ribeiro now had Mr. Davies’ immune cells and even his blood type from the earlier bone marrow transplant, they were a 100 percent match. This also meant she would require fewer antirejection medications.

“It’s the most satisfying thing I’ve ever done,” says Mr. Davies, 38. For her part, Ms. Ribeiro says, “I never met anyone so selfless. He really acted like it was no big deal. But he saved my life twice. He is my guardian angel, and he is now family forever.”

Ms. Ribeiro’s surgeon, Vikram Wadhera, MBBS, Assistant Professor, Surgery, says, “The surgery for both patients went extremely well but, for most of us involved, it was the human and emotional aspects of this case that touched us deeply.”

“I never met anyone so selfless. He really acted like it was no big deal. But he saved my life twice. He is my guardian angel, and he is now family forever.”

—Kelly Ribeiro

Says donor surgeon Edward Chin, MD, Professor of Surgery, and Director of the Living Kidney Donor Program at the Mount Sinai Health System: “This is such a compelling story. It was such an altruistic thing for Jeramy to do. It reminds us that there’s so much good in the world.”

This winter, Ms. Ribeiro has been regaining her strength and looking forward to jump-starting her life, which had been on hold for the past six years. She expects to complete her master’s degree in Library and Information Science from Rutgers University later this year. Mr. Davies, who now lives in Denver, says he is close to getting back to his routine. He is looking forward to snowboarding and training for a triathlon later this year.

Living donation is the shortest route to organ transplantation and often results in a closer match and better outcome for the recipient. The Zweig Family Center for Living Donation at Mount Sinai is one of the largest living donor programs in the United States. For information, please call 212-659-8024.

Mount Sinai Ranked as Top Employer for Diversity

Forbes has ranked the Mount Sinai Health System No. 1 nationally among health systems and hospitals, and No. 19 overall, on its 2019 list of “The Best Employers for Diversity.” The rankings included 500 U.S. organizations in major industries such as biotechnology, insurance, telecommunications, retail, and education.

To compile its second annual diversity list, Forbes partnered with the market research firm Statista to survey 50,000 U.S. employees who work for companies that employ at least 1,000 people nationally. Those surveyed were asked about diversity within their own workplaces and were also given the chance to evaluate employers in their respective industries. Additional criteria that factored into the rankings included diversity among top executives and the board, and leadership that proactively communicated the importance of diversity and inclusion and promoted it throughout their organizations.

“We are thrilled to receive this recognition,” says Gary C. Butts, MD, Chief Diversity and Inclusion Officer for the Mount Sinai Health System, and Dean for Diversity Programs, Policy and Community Affairs for the Icahn School of Medicine at Mount Sinai. “Diversity and inclusion are integral to Mount Sinai’s mission. We are committed to embracing and advancing diversity in our faculty, staff, students, and trainees, and providing our diverse patient population with high-quality care.”

Says Pamela Y. Abner, MPA, CPXP, Vice President and Chief Administrative Officer, Office for Diversity and Inclusion, Mount Sinai Health System, “We strive to create an inclusive and equitable environment where, through the lens of cultural awareness, we engage staff in key diversity and inclusion initiatives to enhance the patient experience.”
More than 200 people recently attended the inaugural NYC Grand Hack at the Icahn School of Medicine at Mount Sinai. The participants came from many institutions, in fields ranging from medicine to programming. At the event, they tackled medical issues in three tracks: rehabilitation and human performance, public and mental health, and lung cancer. The hackathon was organized by a team from the Icahn School of Medicine’s Department of Rehabilitation and Human Performance, along with MIT Hacking Medicine, a group of students and community members from the Massachusetts Institute of Technology that aims to energize and accelerate medical innovation. “It was incredible to watch this extremely diverse group of talented people create innovations,” says an organizer of the event, Salman Hirani, MD, PGY-4, Rehabilitation and Human Performance, Icahn School of Medicine.

Heart health fairs were held throughout the Mount Sinai Health System in honor of Go Red for Women Day* on Friday, February 1. The fairs, which attracted more than 1,000 participants, provided free screenings for blood pressure, total cholesterol, triglyceride levels, and body mass index. The events recognized the American Heart Association’s Go Red campaign, which aims to raise awareness of heart disease, the No. 1 killer of women worldwide. “A shared commitment to the cardiovascular health and well-being of our community makes this a success every year,” says Beth Oliver, DNP, RN, Senior Vice President, Cardiac Services, Mount Sinai Health System.
Personal Nutrition Consultation

Free personal nutrition consultation and diabetes coaching and education will be offered by the Diabetes Alliance.

Mount Sinai Queens Ambulatory Pavilion
2520 30th Avenue, Fourth Floor

Monday, February 25
Tuesday, February 26
Thursdays, February 21 and 28

Email wellness@mountsinai.org to learn about other dates and locations.

Grand Rounds / Geriatrics and Palliative Medicine

Ann-Gel Palermo, DrPH, MPH, Associate Dean, Diversity and Inclusion in Biomedical Education, Associate Professor, Medical Education, and Assistant Professor, Pediatrics, presents “Unconscious Bias Training.”

Thursday, February 21
5 - 6 pm
Annenberg 10-30

Grand Rounds / Obstetrics and Gynecology

Karen Wilson, MD, MPH, the Debra and Leon Black Professor of Pediatrics, Vice Chair for Clinical and Translational Research, Department of Pediatrics, presents “Secondhand Smoke Exposure in Children and Pregnant Women.”

Tuesday, February 26
8 - 9 am
Mount Sinai West, Conference Room 2B

Grand Rounds / Medicine

Pascale White, MD, Director, Gastroenterology Clinic, Assistant Professor, Medicine (Gastroenterology), presents “Colorectal Cancer Screening in African Americans.”

Tuesday, February 26
8:30 - 9:30 am
Hatch Auditorium

Cell, Developmental and Regenerative Biology Seminars

Stacey Ogden, PhD, Associate Professor, Cell and Molecular Biology, St. Jude Children’s Research Hospital, presents “Getting Hedgehog Ligands Where They Need to Go.”

Thursday, February 28
Noon - 1 pm
Annenberg 25-51 Conference Room

“Epic OpTime” Coming to The Mount Sinai Hospital and Mount Sinai Queens

The Epic OpTime Module, an operating room management system for hospitals and ambulatory surgical centers, will launch on Sunday, April 7. It will be followed by the addition of the Epic Anesthesia Module later this year. Adding these modules to Mount Sinai’s Epic electronic medical record system will provide clinicians access to comprehensive patient information, allowing for surgery orders to be managed throughout the perioperative process, beginning with the initial consult, through the day of surgery. The goal is to better coordinate clinical care in real time and improve patient safety through system alerts and tools.

Among the expected benefits:

• Ambulatory surgical centers using Epic Ambulatory will be able to electronically submit surgical case requests.

• Surgeons using Epic Ambulatory will be able to see both their ambulatory appointments and surgical cases in one place.

• One patient record for all Mount Sinai Epic-enabled hospitals and ambulatory practices.

• Integration of documentation with the emergency room, inpatient floors, and ambulatory offices.

Epic OpTime “Go-Live”
Sunday, April 7, early morning

For more information, contact EpicMSHQ_Opt_Anes@mountsinai.org.

Second Annual Live Surgery Training Course For Gender-Affirmation Procedures

This course is presented by The Mount Sinai Hospital and the World Professional Association for Transgender Health (WPATH). The course includes didactic and live gender-affirmation procedures for transgender patients, covering the full range of medical, mental health, and social supports for patients undergoing gender transition.

To register, visit wpath.org/education/upcoming-conferences.

Thursday, February 28 - Saturday, March 2
Stern Auditorium

The Mount Sinai Health System complies with applicable Federal civil rights laws and does not discriminate, exclude, or treat people differently on the basis of race, color, national origin, age, religion, disability, sex, sexual orientation, gender identity, or gender expression.