

Publications of the Week

Mitochondrial Variant Enrichment from High-Throughput Single-Cell RNA Sequencing Resolves Clonal Populations

First Author: Tyler Miller (pictured, right) | Senior Author: Peter van Galen (left) Nature Biotechnology | Massachusetts General Hospital, Broad Institute, Dana-Farber Cancer Institute, Brigham and Women's Hospital, Harvard Medical School, Boston Children's Hospital, and the Koch Institute



The combination of single-cell transcriptomics with mitochondrial DNA variant detection can be used to establish lineage relationships in primary human cells, but current methods are not scalable to interrogate complex tissues. The authors combine common 3' single-cell RNA-sequencing protocols with mitochondrial transcriptome enrichment to increase coverage by more than 50-fold, enabling high-confidence mutation detection. [Profile](#) | [Abstract](#)

Silent Mutations Reveal Therapeutic Vulnerability in RAS Q61Cancers

First Author: Yoshiea Kobayashi | Senior Author: Pasi Jänne (pictured) Nature | Dana-Farber Cancer Institute, Harvard Medical School, and Harvard T. H. Chan School of Public Health



RAS family members are the most frequently mutated oncogenes in human cancers. The authors uncover the requirement of the silent KRAS^{G60G} mutation for cells to produce a functional KRAS(Q61K). In the absence of this G60G mutation in KRAS^{Q61K}, a cryptic splice donor site is formed, promoting alternative splicing and premature protein termination. [Abstract](#)

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Local News

Study Reveals How to Activate Natural Killer Cells to Protect Against Cancer and Other Diseases

Massachusetts General Hospital



New research led by Dr. Shawn Demehri (pictured) at Massachusetts General Hospital reveals factors that control the interplay of natural killer cells — which are part of the body's innate, or first line, immune response — with tumor cells, viral infections, and solid organ transplants. The results could be used to help protect people from cancer, invading pathogens, autoimmunity, inflammatory diseases, and transplant rejection. [Read More](#)

How Autism Risk Genes Converge

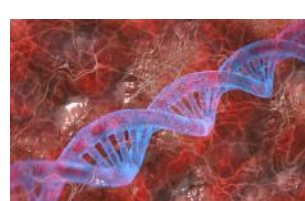
Harvard Brain Science Initiative



Autism spectrum disorder has been recently associated with hundreds of different risk genes, yet it is unclear whether mutations in these genes converge on a similar neurodevelopmental abnormality. Researchers in Dr. Paula Aulotta's (pictured) lab decided to investigate whether three autism-risk genes, *SUV420H1*, *ARID1B*, and *CHD8*, all with very broad hypothetical function, would converge on similar cellular phenotypes. [Read More](#)

Sudden, Unexplained Child Deaths Often Have a Genetic Cause

Boston Children's Hospital



When a baby or toddler dies without warning, parents often blame themselves. A study at Boston Children's may provide some insight into sudden, unexplained child deaths and perhaps a measure of closure. It suggests that at least ten percent of children who die suddenly have an undiagnosed genetic condition that caused or contributed to their death. [Read More](#)

A Potential New Test for Diagnosing Lyme Disease

Tufts University



For scientists and clinicians alike, one of the Holy Grails for successfully treating and curing Lyme disease is developing tests that identify the disease sooner, show when people are cured of infection, and can diagnose reinfection. Now, researchers at Tufts University School of Medicine say they have identified just such a testing mechanism. [Read More](#)

Countering Gut Inflammation

Harvard Medical School



Bile acids made by the liver have long been known for their critical role in helping to absorb the food we ingest. But, according to a series of new studies from Harvard Medical School, these fat- and vitamin-dissolving substances are also important players in gut immunity and inflammation because they regulate the activity of key immune cells linked to a range of inflammatory bowel conditions, such as ulcerative colitis and Crohn's disease. [Read More](#)

Researchers Catalog White Fat Cell Types Present in Mice and in Humans

Broad Institute



Dr. Evan Rosen (pictured) and a team of scientists at Beth Israel Deaconess Medical Center and the Broad Institute of MIT and Harvard have generated an atlas of the cell types present in the white adipose tissue of humans and of mice, allowing them to explore the composition of white fat in unprecedented detail. Their work describes novel subpopulations of fat cells, and links specific cell types to increased risk of metabolic disease. [Read More](#)

Using Genetics to Glimpse Newborns' Future with Congenital Heart Disease

Boston Children's Hospital



Aside from infections, congenital heart disease (CHD) is the leading cause of infant mortality. Dr. Sarah Morton (pictured), Attending Physician in the Division of Newborn Medicine at Boston Children's, has devoted her career to understanding CHD's genetic causes. Her findings are shedding light on what parents and care providers can expect as these babies grow up. [Read More](#)

A Possible New COVID-19 Vaccine Could Be Accessible for More of the World

MIT News



While many people in wealthier countries have been vaccinated against COVID-19, there is still a need for vaccination in much of the world. A new vaccine developed in Drs. J. Christopher Love's (pictured) and Dan Barouch's labs may aid in those efforts, offering an inexpensive, easy-to-store, and effective alternative to RNA vaccines. [Read More](#)

Ana Raquel Santa Maria on Breaking Through the Blood-Brain Barrier to Treat Brain Disease

Wyss Institute



Dr. Ana Raquel Santa Maria (pictured) likes to use the analogy that the brain is like a castle, a fantastic and important place that's being protected by several safeguards, one of which is the blood-brain barrier. She is part of the Wyss Institute's Brain Targeting Program that's working to find a protein that can break through the castle's defenses and deliver drugs for brain diseases where they're needed. [Read More](#)

The Immune System Is Very Complicated, but Now, It's on a Chip

Wyss Institute



Wyss Institute researchers cultured human B and T cells inside a microfluidic organ chip device and coaxed them to spontaneously form functional lymphoid follicles (LFs). "Our LF chip offers a way to model the complex choreography of human immune responses to infection and vaccination, and could significantly speed up the pace and quality of vaccine creation in the future," said Dr. Girija Goyal (pictured). [Read More](#)

An Enzyme's Protective Role

Harvard Medical School



Scientists at Harvard Medical School and Joslin Diabetes Center, led by Dr. George King (pictured), have shed new light on how hyperglycemia contributes to kidney disease, and they have uncovered a potential therapeutic target. Their work rests on a unique, long-running observational study of more than 1,000 people living with type 1 diabetes known as the Medalist Study. [Read More](#)

An "Oracle" for Predicting the Evolution of Gene Regulation

MIT News



Despite the sheer number of genes that each human cell contains, these so-called "coding" DNA sequences comprise just one percent of our entire genome. The remaining 99 percent is made up of "non-coding" DNA — which, unlike coding DNA, does not carry the instructions to build proteins. One vital function of this non-coding DNA, also called "regulatory" DNA, is to help turn genes on and off, controlling how much (if any) of a protein is made. [Read More](#)

Making Steak from Vegetable Protein

Wyss Institute



"Do you like meat?" That was the first question Dr. Kit Parker (pictured) asked Dr. Christophe Chantre when he interviewed for a position in Dr. Parker's Disease Biophysics Group. Dr. Parker was looking for people to work on a project producing meat alternatives from vegetable proteins, using a technology originally designed to create nanofiber wound dressings and body armor. [Read More](#)

Scientists Identify Neurons That Drive Competition and Social Behavior

Massachusetts General Hospital



New research in mice has identified neurons in the brain that influence competitive interactions between individuals and that play a critical role in shaping the social behavior of groups. The findings will be useful not only for scientists interested in human interactions, but also for those who study neurocognitive conditions such as autism spectrum disorder and schizophrenia that are characterized by altered social behavior. [Read More](#)

Genetic Risk Factors for PCOS Act Independently of Ovarian Function

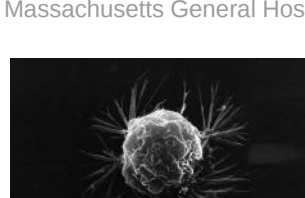
Boston Children's Hospital



It's estimated that as many as ten percent of women have polycystic ovary syndrome (PCOS), a hormonal condition that produces symptoms such as infertility, irregular menstrual periods, and excessive body hair growth. Aiming to learn more about the causes of PCOS, Dr. Jia Zhu (pictured) and her colleagues used a 2018 study on PCOS in women as a springboard to calculate a PCOS polygenic risk score for 176,360 men who shared health data with the U.K. Biobank. [Read More](#)

Lower, More Frequent Doses of Nanomedicines May Enhance Cancer Treatment

Massachusetts General Hospital



Tiny structures called nanoparticles can be used to carry substances to certain parts of the body — for example, to deliver a chemotherapy drug to a tumor. Although such "nanomedicine" offered hope for improving cancer therapeutics, the survival benefits of clinically approved nanomedicines are often modest when compared with conventional chemotherapy. [Read More](#)

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Upcoming Events in Boston

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| March 23 1:00 PM | Finding Funding for Pediatric Medical Research: Meet the Thrasher Research Fund Online |
| March 24 2:00 PM | Biomedical Informatics Entrepreneurs Salon: Diana Brainard, AlloVir Online |
| March 29 5:30 PM | SCIENCE with/in/sight: How Are Cancer Researchers Fighting COVID-19? Koch Institute |
| March 30 11:00 AM | Careers in Academia for Biomedical Students: Panel Online |
| March 31 10:00 AM | 13th Annual Prostate Cancer Awareness Day Online |

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