Department of Pathology

Laboratory Updates

Critical Chromosome Breakage Testing Now Available at BIDMC Pathology Department

The Pathology Department Cytogenetics Laboratory has brought in-house the ability to provide critical chromosome breakage testing for Fanconi Anemia (FA), previously established at Dana-Farber Cancer Institute and Children's Hospital. In June, the lab tested a sample from its first patient. “The introduction of this gold standard diagnostic test for Fanconi anemia in department of pathology, represents a critical advancement in our ability to diagnose this rare devastating disease," said Sean Xu, MD, PhD, director of Molecular Hematopathology. “The initiative reflects a strong collaboration between BIDMC and Dana-Farber Cancer Institute—working together to bring cutting-edge tools directly to patient care. For the hospital, it not only enhances our clinical capabilities but also helps us retain patients by offering a seamless, end-to-end cancer care experience within our system.”

The Pathology Department is one of the few laboratories in the country to have this expertise in diagnosing the disease. The lab includes testing, diagnostics, evaluation, and collected data. Often affecting children and teens, FA is characterized by multiple physical abnormalities, bone marrow failure, and a higher risk of cancer. Normally proteins produced by genes form a kind of cellular "machine" that helps detect and repair damaged DNA in blood stem cells (in bone marrow) and other cells in the body. In FA, DNA repair is slowed, and blood stem cells accumulate damaged DNA and do not survive.

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Laboratory Updates

Critical Chromosome Breakage Testing (cont.)

In addition to testing, the BIDMC FA reference center will also provide both diagnostic and consultative services for new and known patients with FA. Although the test is provided for pediatric patients, the BIDMC Cancer Center will be able to provide extraordinary care and the best therapies to these patients throughout their adult life, including first diagnosis, post treatment, and post transplantation.

Transitioning the FA lab to BIDMC is just one of many testing expansions the Pathology Department has implemented in the past year. “We are continuing to build our Precision Diagnostics program,” said Pathology Department Chair Michael H.A. Roehrl, MD, PhD, MBA. “Every several months we are bringing online new next generation sequencing testing, helping our patients get diagnosed more quickly so they can start treatment sooner.” For more information about the FA lab, contact Annie Cheng or go to the BIDMC Clinical Pathology Cytogenetics page.

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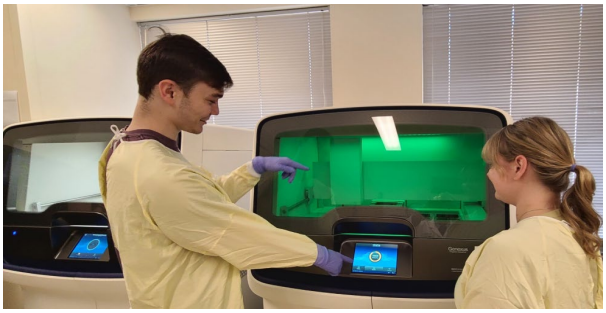
Laboratory Updates

First Next Generation Sequencing Clinical Assay Launched at BIDMC Pathology



Group photo, l-r: Atsushi Tanaka, MD, PhD, pathologist; Darion Tilton, MLS; Megan Pitts, MLS; Tom Moutinho, PhD, Genomic Bioinformatician; Annie Cheng, BSC, ASCP (M, SV), Clinical Manager; Eleanor Kelly, MLS; Samuel Smith, MLS; and Divya Bhagirath, PhD, Lead Genomic Scientist.

As part of the Precision Diagnostics Initiative at the Department of Pathology at BIDMC, spearheaded by the Chair of the Department, Michael H.A. Roehrl, MD, PhD, MBA, the Molecular Pathology Laboratory has developed a Next Generation Sequencing assay designed for rapid and comprehensive analysis of key mutations associated with myeloid malignancies. The Ultra-Fast NGS Myeloid Panel was launched on July 7, 2025, and is already serving the needs of patients of the BIDMC Blood Cancer Program. Leveraging streamlined workflows and optimized bioinformatics pipelines, this assay delivers high-quality results with significantly reduced turnaround time—typically within 3-4 days. It provides critical diagnostic, prognostic, and therapeutic information to support timely clinical decision-making in hematologic oncology. The assay is designed for sensitive detection of myeloid disorder-associated DNA mutations and RNA fusion transcripts in blood and bone marrow samples.

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Laboratory Updates

First Next Generation Sequencing Clinical Assay Launched at BIDMC Pathology (cont.)

This is the first of a series of NGS-based assays to support cancer care at BIDMC. A complete genomic profiling assay, comprised of a large DNA panel and an RNA fusion panel is also under development and is expected to go live at the end of the year. The laboratory has also developed a set of ultra-fast singe-gene assays with a laboratory turnaround time of < 24 hours for common mutations in the *EGFR, KRAS, BRAF*, and *NRAS* genes in solid tumors. Additional ultra-fast assays include an RNA fusion test for solid tumors, *IDH1/IDH2* ingliomas, and *POLE/POLD1* in endometrial cancer. These advancements are supported by the expert faculty at Pathology as well as the clinical oncologist community at BIDMC.

Biomarker Assays

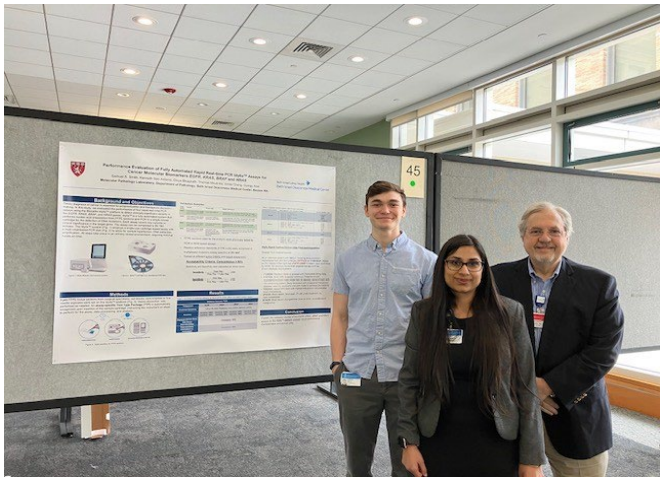
The immunohistochemistry laboratory has recently validated an **MDM2 in-situ hybridization assay**, enabling more rapid and precise detection of MDM2 gene amplification in challenging soft tissue tumors. In addition, our **FOLR1 immunohistochemistry assay** is now available, supporting targeted therapy selection in gynecologic malignancies. We are also nearing completion of validation for **Claudin-18**, a promising biomarker for gastric and gastroesophageal adenocarcinomas with implications for targeted treatments.

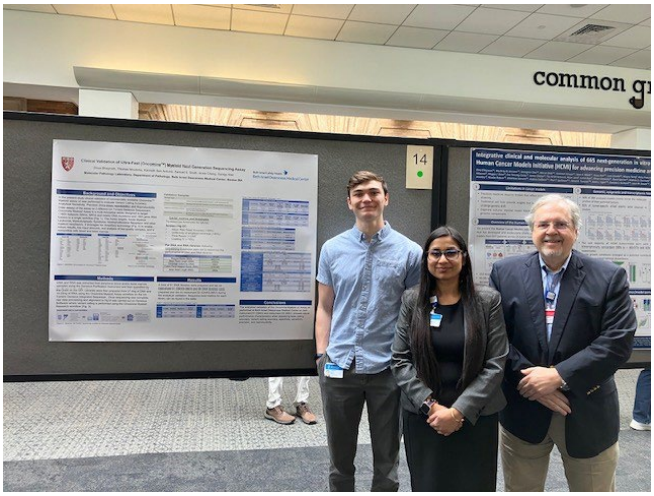
Department of Pathology

News

Harvard Medical School Pathology Research Retreat, May 2025

Congratulations to all the BIDMC pathologists who participated in the 2025 Harvard Medical School Pathology Research Retreat. **Winston Hide, PhD,** presented "Multi-Modal Interrogation of Molecular Mechanisms of Neuronal Resilience and Resistance in Alzheimer's Disease," and **Alex Toker, PhD,** presented "Targeting the PI3K/AKT Pathway in Cancer." The poster session included a dozen BIDMC Pathology submissions. Pictured below, l-r, **Samuel Smith; Divya Bhagirath, PhD; Gyorgy Abel, MD, PhD,** in front of their two posters: "Performance Evaluation of Fully Automated Rapid Real-Time PCR Idylla TM Assays for Cancer Molecular Biomarkers EGFR, KRAS, BRAF, and NRAS" and "Clinical Validation of the Ultra-Fact Oncomine TM Myeloid Next Generation Sequencing Assay."





Department of Pathology

Annual Monroe J. Schlesinger Lecture

Gordon J. Freeman, PhD, Discussed the B7 Gene Family: “A Goldmine of Therapeutic Agents”

Gordon J. Freeman, PhD, Professor of Medicine at Dana Farber Cancer Institute (DFCI) and Harvard Medical School (HMS) presented “B7 Family and Friends: Adventures in Tumor Immunology,” on June 5, as the Monroe J. Schlesinger Lecture, the highlight of the academic year for the BIDMC Department of Pathology. Dr. Freeman called the B7 gene family “a goldmine of therapeutic agents,” which he has been investigating and mining for 38 years. He has been using them to find the optimal T cell expansion for immunotherapy. His work contributes to the groundbreaking approach to cancer treatment that leverages the body’s own immune system to fight cancer cells.

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Schlesinger Lecture (cont.)

The annual lecture is named for Monroe J. Schlesinger, MD who was a distinguished pathologist and the first Chair of the Department of Pathology at Beth Israel Hospital in 1928. Current Pathology Department Chair Michael H.A. Roehrl, MD, PhD, MBA, said of Dr. Schlesinger, “He epitomized the ideals and importance of the investigator physician-scientist.” He added that “named lectures connect us to our past and honor our leaders, past and present.”

Dr. Freeman discussed his research identifying the major pathways that control the immune response by inhibiting or stimulating T cell activation. He has shown that the PD-L1 molecule in the B7 family is highly expressed on many solid tumors, such as breast and lung, as well as some hematologic malignancies. This work provides critical translational insights for developing a successful strategy for cancer immunotherapy. Much of his work targeting molecules for different kinds of cancer is in clinical trials. More recently, he has been investigating how gut bacteria can also be recruited to enhance anti-tumor immunotherapy in mice and people.

He has been an active collaborator with many researchers at BIDMC, including **Rupal Bhatt, MD, PhD**, Hematology/Oncology; **Wenyi Wei, PhD,** Pathology; **David Avigan, MD,** director of the BIDMC Cancer Center; and **David McDermott, MD,** co-director of the BIDMC Immunotherapy institute.

He was hopeful that the new collaboration between DFCI and BIDMC will make more cellular and T cell therapies possible for our patients and ended the lecture with a photo of Barry Nelson who received immunotherapy 12 years ago for what was at that time considered an incurable lung cancer. He is healthy today and an advocate for immunotherapy.

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Schlesinger Lecture (cont.)

“Dr. Freeman’s seminal work in immunology and T cell regulation has paved the way for modern immunotherapy of cancers and other diseases. His laboratory’s work shows how important basic science is for advancing cures for patients, which is also a paradigm for the central role of the medical specialty of Pathology,” said Dr. Roehrl.

Dr. Freeman has published more than 400 papers, three of which have been cited more than 5,000 times, and he is an inventor holding over 98 patents on immunotherapies. He earned his BA in biochemistry and molecular biology, and a PhD in microbiology and molecular genetics from Harvard University. He was elected to the National Academy of Sciences and the National Academy of Inventors and has received numerous awards, including the William B. Coley Award for Distinguished Research in Tumor Immunology, the Warren Alpert Foundation Prize, and the Richard Smalley, MD, Memorial Award.