

Blood Test Could Replace Bone Marrow Biopsies in Multiple Myeloma

Key Takeaways

- SWIFT-seq offers a less invasive alternative to bone marrow biopsies for diagnosing and monitoring multiple myeloma and its precursors, MGUS and SMM.
- The test uses single-cell sequencing to profile circulating tumor cells, providing detailed genetic insights and surpassing traditional methods like FISH.
- In a study, SWIFT-seq accurately detected circulating tumor cells in 90% of patients, showing potential for enhanced risk stratification and genomic monitoring.
- SWIFT-seq's capabilities may influence future drug development by identifying gene signatures that capture tumor circulatory capacity.

SWIFT-seq revolutionizes multiple myeloma diagnosis, offering a non-invasive blood test that enhances monitoring and risk assessment for patients.

SWIFT-seq, a novel blood test developed by researchers from the Dana-Farber Cancer Institute, may change the way multiple myeloma (MM) is diagnosed and monitored, offering patients a less invasive alternative to traditional bone marrow biopsies. Notably, the test may be able to detect the MM precursor diseases monoclonal gammopathy of undetermined significance (MGUS) and smoldering multiple myeloma (SMM).



Gloved hand holding blood sample | Image Credit: ©
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MM is the second most common hematologic malignancy, affecting over 130,000 people in the United States every year. It is a heterogeneous, progressive disease, marked by increasingly poorer treatment responses. MM is preceded by MGUS and SMM—2 premalignant conditions defined by the presence of bone marrow plasma and monoclonal proteins, or M proteins, in the blood. These conditions are typically asymptomatic; however, patients who progress to high-risk SMM may experience symptoms such as fatigue, bone pain, or numbness or tingling in

the hands and feet.¹⁻³

Diagnosis, risk assessment, and monitoring of MGUS, SMM, and MM require a bone marrow biopsy, which can be painful, infrequent, and lack the accuracy needed for proper patient disease screening. This testing method relies on FISH, or fluorescence in situ hybridization, which often fails to provide clear test results. In response to this critical need for an advanced, less invasive testing option, researchers explored the potential of profiling circulating tumor cells (CTCs).⁴

“BM biopsies are conducted infrequently and can yield inconclusive results due to technical limitations,” the authors wrote. “Profiling [CTCs] may enable noninvasive routine clinical assessments but remains challenging.”⁵

SWIFT-seq is a blood test that utilizes single-cell sequencing to profile CTCs, allowing doctors to perform risk assessments and genetic monitoring using a liquid biopsy test. This makes the process easier for both patients and providers, as well as offering more detailed, accurate measurements for disease.⁴

SWIFT-seq provides a detailed genetic profile beyond counting CTCs to help identify key genetic changes crucial for understanding MM, offering superior accuracy to traditional bone marrow tests like FISH. SWIFT-seq is capable of assessing tumor growth rates as well as identifying gene patterns to project patient outcomes.⁴

“SWIFT-seq is a powerful option, as it can measure the number of CTCs, characterize the genomic alterations of the tumor, estimate the tumor’s proliferative capacity, and measure prognostically useful gene signatures in a single test and from a blood sample,” Irene M. Ghobrial, MD, senior author, said in a news release.⁴

The capabilities of SWIFT-seq were observed in a study of 101 patients with MGUS, SMM, or MM, and healthy donors. The authors reported that SWIFT-seq accurately detected CTCs in 90% of patients with MGUS, SMM, and MM. CTCs were detected in 95% of patients with SMM and 94% of those with newly diagnosed MM, populations most likely to benefit from enhanced risk stratification and genomic monitoring.⁴

“We identified a gene signature that we believe captures the tumor’s circulatory capacity and may partly explain some of the unexplained mysteries of myeloma biology,” Elizabeth D. Lightbody, MD, co-first author, said in the news release. “This can have a tremendous impact on how we think about curtailing tumor spread in patients with myeloma and could lead to the development of new drugs for patients.”⁴

REFERENCES

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