Rarity Bioscience Launches Ultra-Sensitive KIT D816V Assay, a New Benchmark for Molecular Testing in Systemic Mastocytosis

Innovative superRCA® assay enables quantitative KIT D816V detection down to less than 0.001% variant allele frequency

Uppsala, Sweden — September 17th 2026 — **Rarity Bioscience** today announced the launch of the breakthrough superRCA Ultra-Sensitive KIT D816V Kit RUO, an ultra-sensitive assay for quantitative detection of the *KIT* D816V mutation. Developed in close collaboration with leading experts from Blueprint Medicines, a Sanofi company, and internationally recognized specialists in systemic mastocytosis (SM), the assay enables accurate and quantitative detection of *KIT* D816V at variant allele frequencies less than 0.001%, establishing a new benchmark for sensitive mutation detection.

KIT D816V is the primary driver mutation in SM and may be present at very low levels, particularly in peripheral blood, making ultra-sensitive detection essential. By enabling reliable quantification of trace amounts of mutant DNA, the superRCA ultra-sensitive KIT D816V assay expands the utility of peripheral blood to advance research into disease biology and clinical diagnostics.

"This launch is more than the introduction of a new assay. It reflects our broader vision for the future of molecular analysis," said Linus Bosaeus, CEO of Rarity Bioscience. "We believe that the next generation of precision medicine will depend on technologies capable of detecting the rarest disease signals with exceptional accuracy while remaining accessible to laboratories worldwide. By continuing to expand the capabilities of the superRCA® platform, we aim to help researchers unlock new biological insights and accelerate the development of better diagnostics and therapies."

The assay was developed through an ongoing collaboration with leading experts from Blueprint Medicines, a Sanofi company, as well as internationally recognized clinicians and researchers in SM. Their expertise helped optimize the assay for the practical clinical challenges of detecting and quantifying low-level *KIT* D816V mutations, aligning its performance with the needs of patients.

The superRCA ultra-sensitive KIT D816V kit is intended for Research Use Only and will be commercially available in October 2026.

Overcoming Diagnostic Barriers in Systemic Mastocytosis

SM is a rare disease that often leads to debilitating symptoms including skin lesions, diarrhea, fatigue, bone pain and anaphylaxis. The heterogeneity and non-specific nature of SM symptoms can contribute to diagnostic delays; a previous study reported a mean time from symptom onset to diagnosis of about six years.[1] These barriers highlight the need to pursue further research into novel diagnostic approaches.

"Many patients with systemic mastocytosis experience a substantial symptom burden, even when circulating KIT D816V levels are low, underscoring the importance of timely diagnosis and care," said Ben Lampson, M.D., Ph.D., Senior Medical Director of Clinical Research at Blueprint Medicines, a Sanofi company. "Working closely with Rarity Bioscience and clinical research laboratories, we aim to accelerate the development of non-invasive, blood-based tests that enhance detection of KIT D816V. These efforts are part of our long-term commitment to partner with diagnostic experts, clinicians and patient advocates to transform care for people living with SM."

Built on Rarity Bioscience's proprietary superRCA® technology, the assay combines exceptional analytical sensitivity with highly specific mutation detection and robust quantitative performance in a streamlined workflow. By leveraging readily available flow cytometers, it eliminates the need for capital investments, supporting decentralized adoption.

About Rarity Bioscience

Rarity Bioscience is a Swedish biotechnology company pioneering ultra-sensitive nucleic acid detection technologies. Its proprietary superRCA® platform enables high-precision genetic analysis, supporting applications in early disease detection, therapy monitoring, and clinical research.

[1] Mesa RA, Sullivan EM, Dubinski D et al. Patient-reported outcomes among patients with systemic mastocytosis in routine clinical practice: Results of the TouchStone SM Patient Survey. *Cancer*. 2022;128(20):3691-3699.

Media Contact
Loora Laan, Product Manager
loora.laan@raritybioscience.com
www.raritybioscience.com