



PerkinElmer Expands Genomic Testing Services with Ultrarapid Whole Genome Sequencing

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Latest addition to Company's genomic testing services menu delivers rapid results to physicians that inform clinical management and outcomes for critically ill patients

WALTHAM, Mass. – May 16, 2022 – [PerkinElmer, Inc.](#), a global leader committed to innovating for a healthier world, today announced the availability of [ultrarapid whole genome sequencing](#) (urWGS) through [PerkinElmer Genomics](#). This addition to the Company's portfolio of whole genome sequencing (WGS) offerings provides physicians with comprehensive, meaningful results in five days to help inform clinical management and improve outcomes for critically ill patients in neonatal and pediatric intensive care units (NICUs and PICUs). With many genetic diseases being chronic and progressive in nature, reducing the time to reaching an accurate diagnosis can eliminate unnecessary procedures, initiate treatment and improve clinical outcomes.

The PerkinElmer Genomics urWGS offering uses a dried blood spot sample to provide phenotype-driven analysis with a mean coverage of 40x of a patient's genome. Studies suggest that [up to 15 percent](#) of disease-causing genetic variants may be found in the non-coding regions of the genome, which WGS helps identify. In addition to an analysis of the mitochondrial genome, CNV detection – which identifies deletions, duplications, and other gene- and chromosomal-level events, SMA and a repeat disorders screen – the PerkinElmer urWGS offering includes a [StepOne® Comprehensive Biochemical Profile](#). The StepOne offering screens for more than 70 inherited conditions and disorders, including the Recommended Universal Newborn Screening Panel (RUSP) and many others that may not be found in state-mandated programs.

"Increasingly, whole genome sequencing is proving its value as a first-tier clinical test for many patients, especially in a NICU and PICU where timely clinical decisions are critical for timely intervention," said Madhuri Hegde, PhD, FACMG, SVP and chief scientific officer, Global Lab Services, PerkinElmer Inc. "By making services such as this urWGS combined with StepOne available to more hospital systems and physicians, our hope is that we may help shorten the diagnostic odyssey for more newborns and their families. With PerkinElmer's global leadership in newborn screening using dried blood spot card technology, we will continue to enhance urWGS with additional assays."

As a result of informing and initiating changes in clinical management, rapid WGS tests [have been shown to reduce healthcare costs](#) for patients in NICUs and PICUs. Additional benefits of these testing services include the elimination of unnecessary tests and procedures, and reduced length of hospital stays.

PerkinElmer Genomics is among the first commercial clinical laboratories to receive approval from the State of New York for its next generation sequencing-based method for WGS using saliva, whole blood, and dried blood spot specimens. In December 2021, the Company also launched a [prenatal WGS test](#) offering, further expanding its capabilities as a leader in this space.

The PerkinElmer Genomics urWGS test is offered as a proband-only or TRIO test. For more information on both tests, ordering instructions and a summary of all PerkinElmer Genomics WGS offerings, visit [the webpage here](#).

About PerkinElmer

PerkinElmer is a leading, global provider of end-to-end solutions that help scientists, researchers and clinicians better diagnose disease, discover new and more personalized drugs, monitor the safety and quality of our food, and drive environmental and applied analysis excellence. With an 85-year legacy of advancing science and a mission of innovating for a healthier world, our dedicated team of more than 16,000 collaborates closely with commercial, government, academic and healthcare customers to deliver reagents, assays, instruments, automation, informatics and strategic services that accelerate workflows, deliver actionable insights and support improved decision making. We are also deeply committed to good corporate citizenship through our dynamic ESG and sustainability programs. The Company reported revenues of approximately \$5 billion in 2021, serves customers in 190 countries, and is a component of the S&P 500 index. Additional information is available at www.perkinelmer.com. Follow PerkinElmer on [LinkedIn](#), [Twitter](#), [Facebook](#), [Instagram](#), and [YouTube](#).

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