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**PERSONAL GENOME DIAGNOSTICS' CIRCULATING TUMOR DNA TECHNOLOGY HIGHLIGHTED  
IN LANDMARK STUDY**

**—New Publication in *Science Translational Medicine* Confirms Utility of Cell-Free Circulating Tumor DNA (ctDNA) for Cancer Drug Development and Diagnosis—**

**—Personal Genome Diagnostics Is Providing ctDNA Genomic Testing to Cancer Drug Developers Using Its Proprietary PARE and Related Technologies—**

**BALTIMORE, MD, February 19, 2014** – Personal Genome Diagnostics Inc. (PGDx), a provider of advanced cancer genome analysis and testing services, reported that its proprietary technology was used in a major new study being published today in the journal *Science Translational Medicine*. The study, “Detection of Circulating Tumor DNA in Early- and Late-Stage Human Malignancies,” assessed the utility of cell-free circulating tumor DNA (ctDNA) for cancer detection and monitoring across multiple patient populations and applications. The authors conclude that ctDNA is a broadly applicable, sensitive and specific biomarker that can be used for a variety of clinical and research purposes in patients with different types of cancer.<sup>1</sup>

Some of the findings reported today were analyzed using PGDx's proprietary PARE technology, an ultrasensitive technique that enables whole genome identification of changes in tumor-specific ctDNA. The new findings are consistent with previous publications by PGDx researchers and further validate the company's work using ctDNA-based analyses for cancer research and drug development.

“This landmark study further validates the utility of circulating DNA as a biomarker in cancer therapeutic research and treatment,” noted Mark Sausen, PhD, a lead author of the new study and Director of R&D at PGDx. Dr. Sausen was one of the first researchers to use genome-wide analyses of ctDNA to identify newly-acquired genetic alterations associated with resistance to targeted therapies.

Dr. Sausen added, “At PGDx, we have experienced a surge of interest in ctDNA-based analyses and are now using our PARE and related technologies to routinely perform genetic analyses on ctDNA, thereby obviating the need for invasive biopsies. Genomic analyses conducted using ctDNA produce a great deal of information, including detection of structural genomic alterations and point mutations, which makes them very valuable for the development of targeted cancer therapies.”

Unlike other approaches, which can detect point mutations in ctDNA, PARE (Personalized Analysis of Rearranged Ends) can also detect structural changes, including the genomic amplifications and rearrangements that are critical for guiding cancer treatment. PGDx has licensed exclusive rights to the PARE technology from Johns Hopkins University.

The study in *Science Translational Medicine* addressed several topics related to ctDNA. The first assessed the extent to which ctDNA could be detected in different cancer populations at various stages of disease. It found ctDNA was detectable in many, but not all types of advanced solid tumor cancers, as well as in many individuals with earlier-stage localized tumors. The study also found that cell-free ctDNA often was detectable in patients who lacked circulating tumor cells, providing further validation of PGDx's ctDNA approach.

The new study also found that ctDNA could be used to identify known and novel genetic mutations associated with drug resistance in patients with colorectal cancer. PGDx has previously published research showing that its PARE ctDNA approach<sup>2,3</sup>, which can detect structural alterations in ctDNA with high sensitivity and specificity, was able to identify novel mechanisms of acquired drug resistance from the blood of a colorectal cancer patient being treated with targeted therapy. These analyses enabled the new

genetic alteration to be identified before any clinical signs of drug resistance were evident, providing early warning that a change in therapy was needed.

Antony Newton, Chief Commercial Officer of PGDx, added, “This new study confirms the value of our expertise in the application of cell-free circulating tumor DNA technologies. We are already offering these technologies for both prospective and retrospective clinical trial applications and expect this new study will accelerate the growing interest in the use of ctDNA for drug development.”

Along with Dr. Sausen, co-authors of the new study include PGDx co-founders and Johns Hopkins cancer researchers Dr. Luis Diaz and Dr. Victor Velculescu.

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2. *Oncotarget*. 2013 October; 4(10): 1856–1857. Published online 2013 October 8. PMID: PMC3858570. **Insights into therapeutic resistance from whole-genome analyses of circulating tumor DNA**. Luis A. Diaz, Jr., Mark Sausen, George A. Fisher, and Victor E. Velculescu.

3. *Cancer Discovery*. 2013 June; 3 (6): 658-73. doi: 10.1158/2159-8290.CD-12-0558. **Amplification of the MET receptor drives resistance to anti-EGFR therapies in colorectal cancer**. Bardelli A, Corso S, Bertotti A, Hobor S, Valtorta E, Siravegna G, Sartore-Bianchi A, Scala E, Cassingena A, Zecchin D, Apicella M, Migliardi G, Galimi F, Lauricella C, Zanon C, Perera T, Veronese S, Corti G, Amatu A, Gambacorta M, Diaz LA Jr, Sausen M, Velculescu VE, Comoglio P, Trusolino L, Di Nicolantonio F, Giordano S, Siena S.

### **About Personal Genome Diagnostics**

Personal Genome Diagnostics (PGDx) provides advanced cancer genome analyses to oncology researchers, drug developers, clinicians and patients. The company uses advanced genomic methods and its deep expertise in cancer biology to identify and characterize the unique genomic alterations in tumors. PGDx’s proprietary methods for genome sequencing and analysis are complemented by its extensive experience in cancer genomics and clinical oncology. The founders of PGDx, Luis Diaz, MD, and Victor Velculescu, MD, PhD, are internationally recognized leaders in cancer genomics at Johns Hopkins University who have extensive experience in the practical application of advanced genomic technologies to drug development and clinical practice. PGDx’s CLIA-certified facility provides personalized cancer genome analyses to patients and their physicians. For more information, visit [www.personalgenome.com](http://www.personalgenome.com).

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