



## FIRST DISCOVERY OF ACTIONABLE GENETIC ALTERATIONS IN A DEADLY CANCER REPORTED BY PERSONAL GENOME DIAGNOSTICS AND BLUEPRINT MEDICINES

—*Genomic Data in Study Published in Nature Communications Suggests New Treatment Strategies for These Hard to Treat Women’s Reproductive Cancers*—

**BALTIMORE, MD, and CAMBRIDGE, MA, September 19, 2014** – [Personal Genome Diagnostics](#) Inc. (PGDx), a provider of advanced cancer genome analysis and testing services, and [Blueprint Medicines](#), a leader in discovering and developing highly selective kinase inhibitors for genomically defined cancers, today disclosed the first-ever comprehensive genomic study of malignant mixed Müllerian tumors (MMMT), an aggressive and deadly cancer of the female reproductive system, also known as carcinosarcoma. The new [study](#), published in the current online edition of *Nature Communications*, uncovers genetic alterations previously not associated with MMMT.<sup>1</sup> These genetic alterations likely play a role in development of the tumors and can serve as targets for anticancer drugs.

Researchers from PGDx and Blueprint Medicines, with colleagues from Johns Hopkins University and Oregon Health and Science University, discovered that MMMT/carcinosarcomas have many mutations located in clinically relevant genes, such as *PIK3CA*, *KRAS* and DNA repair pathway genes. Some of these previously unidentified genetic mutations may be addressed by existing therapies or by investigational drugs currently in clinical trials.

Siân Jones, PhD, a co-first author of the study and Director of Genomic Analysis at PGDx, commented, “By defining the mutational landscape of this understudied cancer with a poor prognosis, PGDx and Blueprint Medicines were able to identify alterations in specific genes and pathways that may be promising targets for existing and new drug therapies, as well as enabling earlier and more effective diagnoses. It is noteworthy that many of the genetic alterations we identified were not previously associated with MMMT/carcinosarcomas. These findings show how comprehensive genomic analysis of a complex type of cancer can increase understanding of the condition, identify potential new treatment options and enable personalized patient management.”

The analysis also revealed that a high fraction of mutations were in “chromatin remodeling” genes, which regulate the structure of chromosomes and, when mutated, are thought to have dramatic effects on the biology of the cell. Chromatin remodeling genes are currently being evaluated as potential targets for epigenetic and other novel therapies.

“The collaborative work between Blueprint Medicines and PGDx demonstrates our resolute commitment to uncovering the genomic drivers of underserved cancers and sharing this information broadly to improve cancer research, diagnosis and treatment for the benefit of patients,” said Christoph Lengauer, PhD, MBA, Chief Scientific Officer of Blueprint Medicines. “Our research further shows the potential of the Blueprint and PGDx platforms to develop innovative genomics-based techniques and tools to elucidate novel genomic drivers of cancer for drug discovery purposes.”

Authors of the study include PGDx co-founders and Johns Hopkins cancer researchers Dr. Victor Velculescu and Dr. Luis Diaz, and Blueprint Medicines' Chief Scientific Officer Dr. Lengauer.

1. Genomic analyses of gynaecologic carcinosarcomas reveal frequent mutations in chromatin remodelling genes, Sian Jones, Nicolas Stransky, Christine L. McCord, Ethan Cerami, James Lagowski, Devon Kelly, Samuel V. Angiuoli, Mark Sausen, Lisa Kann, Manish Shukla, Rosemary Makar, Laura D. Wood, Luis A. Diaz Jr, Christoph Lengauer & Victor E. Velculescu, *Nature Communications*, Sept. 19, 2014, <http://dx.doi.org/10.1038/ncomms6006>

### **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).

### **About Blueprint Medicines**

Blueprint Medicines is a patient-driven oncology company discovering and developing highly selective kinase inhibitors for genomically defined cancers. Led by a management team and advisors with world renowned expertise in cancer genomics, drug discovery and clinical oncology, Blueprint Medicines has developed a platform that combines genomics with a novel small molecule library of kinase inhibitors, enabling Blueprint Medicines to rapidly discover potent and highly selective drugs against clear drivers of diseases. Founded in 2011, Blueprint Medicines is privately held and initially backed by Third Rock Ventures and Fidelity BioSciences. For more information, please visit [www.BlueprintMedicines.com](http://www.BlueprintMedicines.com).

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