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As if by magic: Team develops program that lights up cancer-causing mutations

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By conjuring the spell "Lumos!" wizards in the mythical world of Harry Potter could light up the tip of their magic wands and illuminate their surroundings. So, too, does LumosVar, a computer program developed by the Translational Genomics Research Institute (TGen), "light up" cancer-causing genetic Var-ients, or mutations, illuminating how physicians might best treat their patients.

A study published today in the scientific journal *Frontiers in Oncology* describes how researchers at TGen, an affiliate of City of Hope, developed LumosVar to create a tool that can accurately identify cancer-causing mutations from patient tumor samples.

In the case of archived samples from patients for which treatment outcome results are known, these represent a treasure trove of information that could accelerate research by investigators and physicians in predicting responses of future patients to particular treatments.

"There are many open questions in precision oncology that can only be answered by collecting large amounts of patient genomic data linked to treatment response and clinical outcomes," said Dr. Rebecca Halperin, a Research Assistant Professor in TGen's Quantitative Medicine and Systems Biology Program.

"The approach we outline in this study should enable researchers to use archival samples more effectively. Accurately calling, or identifying,

somatic variants—those DNA changes specific to a patient's cancer—are the first step in any analysis," said Dr. Halperin, the study's lead author.

However, archived tumor samples are frequently not accompanied by the patients' normal—or germline—genetic information, making it difficult to distinguish the patient's normal DNA variants to their mutated and cancerous DNA changes.

LumosVar is a precise enough tool that it not only can detect the cancerous DNA from a patient sample, but it also can differentiate the adjacent normal DNA that may surround the tumor in the sample. Comparing the patient's normal DNA from a suspected cancer-causing mutation is critical to eliminating benign, non-cancerous variants in the sample—"false positives"—and ensuring that the tissue sample analysis is as accurate as possible.

A high level of accuracy is needed for physicians to use this information in precision medicine, determining what treatment each individual patient should receive.

"The sequencing of DNA from tissue adjacent to the tumor could help identify somatic, or cancer-causing, mutations when another source of normal tissue is not available," said Dr. Sara Byron, Research Assistant Professor in TGen's Integrated Cancer Genomics Division, and also the study's senior author.

This study—Leveraging Spatial Variation in Tumor Purity for Improved Somatic Variant Calling of Archival Tumor Only Samples—was funded by The Ben and Catherine Ivy Foundation, GE Global Research, and the Texas A&M System Chancellor's Research Initiative.

More information: Rebecca F. Halperin et al, Leveraging Spatial Variation in Tumor Purity for Improved Somatic Variant Calling of

Archival Tumor Only Samples, *Frontiers in Oncology* (2019). DOI: 10.3389/fonc.2019.00119