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Researchers develop novel system for cataloging cancer gene variants

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The discovery of variations in genes in tumor samples has been critical to the understanding of how cancer develops and spreads, and how to effectively treat it. Now, a multi-institutional group of researchers from the National Human Genome Research Institute-supported Clinical Genome Resource, known as ClinGen, including researchers from Baylor College of Medicine and Texas Children's Hospital, have developed a standard way to catalog gene mutations in cancers in order to enhance the use of the information in research and clinical practice.

The result is the minimal variant level data, or MVLD, a uniform cataloging system that presents a standard way for researchers everywhere to refer to relevant data and information about each variant found in cancer studies. A paper published online in *Genome Medicine* describes the cataloging system.

There are many different teams of cancer researchers and clinical laboratories that provide information online and in other databases about gene variations associated with cancer. Two of the larger efforts to catalog cancer variation include Clinical Interpretations of Variants in Cancer (CIViC) and ClinVar, a large database supported by the National Center for Biotechnology Information. However, each group has its own guidelines and presents information in different versions. If this new system is adopted by researchers, it will help to unify the cataloging efforts across databases.

"By establishing the minimal variant level data with a standard format or

language, it will be easier for researchers and the public to use information associated with mutations in cancers," said Dr. Deborah Ritter, research scientist in the Department of Pediatrics at Baylor and co-first author of the paper. "We expect this will be a major contribution to save time and resources."

The minimal variant level data includes common information collected about a genetic change found in a tumor, including what chromosome it is on, whether it is within a gene and whether it was seen only in tumors or also in normal samples. In addition, each genetic change also is characterized by how it might impact the cancer or treatment of the patient where it is found. Developing a standard to present this information was the main focus of the project, Ritter said. Fields within the minimal variant level data cataloging system that present this information are:

- Cancer Type – the type of cancer in which the mutation was seen
- Biomarker Class – whether the mutation is diagnostic, prognostic or predictive of cancer
- Therapeutic Context – which medications have been associated with the mutation
- Effect – response to medications or other treatments for cancers with this mutation
- Level of Evidence – an overall summary statement about how much is known about the mutation and what type of information is available

The driving force behind the creation of this cataloging system was the ClinGen Somatic Working Group co-chaired by Dr. Subha Madhavan of Georgetown University, senior author of the paper. ClinGen is a large, international consortia of academic and industry leaders dedicated to curating clinically important changes in the human genome. Baylor is one of the ClinGen grant recipients under Dr. Sharon Plon, professor of

pediatrics and of molecular and human genetics.

Members of the working group assembled representatives from various institutions and groups and reviewed existing databases to develop the standard. It has been adopted by CIViC, and leaders of the project are working with other cancer organizations and testing laboratories to adopt this new cataloging system.

"We are already using this new standard to make cancer data easier to understand and share with other clinical laboratories and researchers," said Plon.

More information: Deborah I. Ritter et al. Somatic cancer variant curation and harmonization through consensus minimum variant level data, *Genome Medicine* (2016). DOI: 10.1186/s13073-016-0367-z

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