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CIViC: Bridging the Gap between Clinical Treatment and Cancer Research

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Precision, or personalized, medicine seeks to tailor clinical treatment based on each person's genomic makeup. In the context of cancer, this often involves modifying disease treatment based on the presence and absence of known cancer-causing mutations. The biomedical literature describing these associations is huge. But, currently, the publications explaining genotype-phenotype correlations exist largely in private or encumbered databases resulting in extensive repetition of effort and lack of public access to clinically-actionable data, which in turn reduces the effectiveness of precision medicine. Realizing the potential power of precision medicine requires this information be centralized, vetted and interpreted for clinical application and public access.

The Clinical Interpretations of Variants in Cancer (CIViC) project aims to enable precision medicine by providing an open access knowledgebase for clinicians, researchers, and patients to input submission data, vet this information, and learn about the clinical significance of cancer genome alterations.

My project focusing on curating information on the VHL gene, which, when mutated, causes von Hippel-Lindau disease, is underway. Von Hippel-Lindau disease (VHLD) is a rare, autosomal dominant disease affecting 1 in 36,000 people worldwide and manifests as hemangioblastomas of the central nervous system and retina, renal cell carcinomas, and pheochromocytomas. I seek to curate a large and growing dataset on VHL gene alterations, VHLD, and VHLD patient outcomes in order to link actionable genetic data to clinical practice. Specifically using the CIViC platform, I will summarize patient genotypes, their related phenotypes, and any findings for better prognosis and/or diagnosis of VHLD for each available publication.

From the curation of this information, we hope to create a more comprehensive and readily accessible understanding of the clinical relevance of VHL gene variants and their impact on disease phenotype and treatment, taking us a step closer to realizing the power of precision medicine.