

Cancer encompasses a wide range of diseases that can affect any part of the body characterized by the rapid growth of abnormal cells. Based on the World Health Organization, cancer is a leading cause of death worldwide, accounting for nearly 10 million deaths in 2020, or nearly one in six deaths. The significance of early cancer diagnosis cannot be overstated. It often makes the difference between successful treatment and a challenging prognosis. To enable such timely detection and make informed decisions about the risks and benefits of different treatment options, efficient risk prediction algorithms are essential.

Cancer's core lies in a handful of critical genes known as cancer drivers. While genomic studies have unveiled thousands of mutations in cancer driver genes, they mostly appear sporadic and rare. Our intriguing hypothesis suggests that functional alterations required for malignancy may result from a combination of germline genomic changes (inherited genes) and acquired somatic alterations (lifetime mutations) in much bigger number of cancer-related genes. This theory suggests that individuals born with numerous harmful germline variants may acquire critical mutations faster. Our first model, CanAge, leverages genetic data to predict when a specific cancer might develop, offering personalized insights into germline genomic contributions to cancer risk. It may also serve as a foundation for future integrated scores considering both genomic and environmental factors, helping identify individuals with increased cancer development risks.

Imagine a free, publicly available decision support tool that empowers both patients and physicians to make well-informed shared decisions regarding the risks and benefits of cancer treatment. Such a tool could reduce decision-related stress for patients and provide physicians with more objective recommendations. This is where our second model CanSurv comes into play. CanSurv is designed to be a multivariable risk prediction model capable of estimating the breast cancer survival for individual patients and assessing the incremental benefits of any adjuvant therapies for which clinical trial data are available.

Ultimately, the methodologies developed in this project could have a profound impact on personalized medicine in cancer treatment. Personalized medicine aims to understand the unique genetic characteristics of each patient's cancer, allowing for tailored treatment decisions. By comprehending these molecular signatures, healthcare providers can make more informed choices, and patients can receive personalized guidance, enhancing their quality of life.