A simple blood test could lead to better cancer treatment

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Video Abstract

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Abstract

Researchers have taken a significant step towards personalizing the treatment of cancer. Using DNA sequencing, they've developed a way to scan blood samples for mutations in circulating tumor DNA – small bits of genetic material that are released as cancer cells die. The genomic reservoir contained in this DNA is representative of nearly all tumors carried by a patient, providing the foundation needed for comprehensive genetic profiling. Such profiling may help clinicians select the most appropriate therapies for a given patient. The genetic mutations giving rise todrivingthat drive cancer development often differ markedly among individuals. Interventions that match a patient's unique genetic profile offer great promise, but obtaining tumor tissue for genetic testing can be invasive, and risky and sometimes impossible. To bypass these limitations, the researchers established a way to enrichextract, sequence, and analyze circulating tumor DNA. The method, which relies on the targeted and simultaneous sequencing of a panel of 382 cancer-relevant genes simultaneously, is more time and cost effective and faster than traditional whole-genome sequencingsingle- gene testanalysis. The team used the approach to assess 605 patients with 29 different types of solid tumors. The patients were divided into two groups. In the first group, the researchers compared mutation profiles between circulating tumor DNA and actual tumor specimens collected fromin the same patients. Approximately 80% of these patients had abnormalities in circulating tumor DNA, and nearly 75% of the abnormalities matched those found in the tumor tissues. For In the second group, withthe team tested validation purposes, only circulating tumor DNA. was available for tested intesting the second group, . TtThe two groups showedhe results showed similar numbers and types and spectra of mutations to those found in group 1one, suggesting that circulating tumor DNA is a viable alternative for genetic profiling. Even more encouragingly, among all the patients, 71% of the circulating tumor DNA samples contained at least one clinically actionable mutation, and 66% of these could be targeted by drugs already approved or currently in clinical trials. These findings support that a simple blood test can help guide the personalized treatment of cancer. Because of the non-invasive nature of the testing, it can also be easily repeated. This may allow clinicians to better monitor disease progression, potentially getting patients the care they need faster.