

Computer program developed to diagnose and locate cancer from a blood sample

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Researchers in the United States have developed a computer program that can simultaneously detect cancer and identify where in the body the cancer is located, from a patient's blood sample. The program is described in research published this week in the open access journal *Genome Biology*.

Professor Jasmine Zhou, co-lead author from the University of California at Los Angeles, said: "Non-invasive diagnosis of cancer is important, as it allows the early diagnosis of cancer, and the earlier the cancer is caught, the higher chance a patient has of beating the disease. We have developed a computer-driven test that can detect cancer, and also identify the type of cancer, from a single <u>blood sample</u>. The technology is in its infancy and requires further validation, but the potential benefits to <u>patients</u> are huge."

The <u>program</u> works by looking for specific molecular patterns in cancer DNA that is free flowing in the patients' <u>blood</u> and comparing the patterns against a database of tumour epigenetics, from different cancer types, collated by the authors. DNA from tumour cells is known to end up in the bloodstream in the earliest stages of cancer so offers a unique target for early detection of the disease.

Professor Zhou explained: "We built a database of epigenetic markers, specifically methylation patterns, which are common across many types of cancer and also specific to cancers originating from specific tissue, such as the lung or liver. We also compiled the same 'molecular



footprint' for non-cancerous samples so we had a baseline footprint to compare the cancer samples against. These markers can be used to deconvolute the DNA found freely in the blood into tumor DNA and non-tumor DNA."

In this study, the new computer program and two other methods (called Random Forest and Support Vector Machine) were tested with blood samples from 29 liver cancer patients, 12 <u>lung cancer patients</u> and 5 <u>breast cancer patients</u>. Tests were run 10 times on each <u>sample</u> to validate the results. The Random Forest and Support Vector Machine methods had an overall error rate (the chance that the test produces a false positive) of 0.646 and 0.604 respectively, while the new program obtained a lower error rate of 0.265.

Twenty-five out of the 29 liver cancer patients and 5 out of 12 lung cancer patients tested in this study had early stage cancers, which the program was able to detect in 80% of cases. Although the level of tumour DNA present in the blood is much lower during the early stages of these cancers, the program was still able to make a diagnosis demonstrating the potential of this method for the <u>early detection</u> of cancer, according to the researchers.

Professor Zhou added: "Owing to the limited number of blood samples, the results of this study are evaluated only on three <u>cancer</u> types (breast, liver and lung). In general, the higher the fraction of tumor DNAs in blood, the more accurate the program was at producing a diagnostic result. Therefore, tumors in well-circulated organs, such as the liver or lungs are easier to diagnose early using this approach, than in less-circulated organs such as the breast."

More information: Shuli Kang et al, CancerLocator: non-invasive cancer diagnosis and tissue-of-origin prediction using methylation profiles of cell-free DNA, *Genome Biology* (2017). DOI:



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