

A new approach to detecting cancer earlier from blood tests: study

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Cancer scientists led by principal investigator Dr. Daniel De Carvalho at Princess Margaret Cancer Centre have combined "liquid biopsy", epigenetic alterations and machine learning to develop a blood test to

detect and classify cancer at its earliest stages.

The findings, published online today in *Nature*, describe not only a way to detect [cancer](#), but hold promise of being able to find it earlier when it is more easily treated and long before symptoms ever appear, says Dr. De Carvalho, Senior Scientist at the cancer centre, University Health Network.

"We are very excited at this stage," says Dr. De Carvalho. "A major problem in cancer is how to detect it early. It has been a 'needle in the haystack' problem of how to find that one-in-a-billion cancer-specific mutation in the [blood](#), especially at earlier stages, where the amount of tumour DNA in the blood is minimal."

By profiling [epigenetic alterations](#) instead of mutations, the team was able to identify thousands of modifications unique to each cancer type. Then, using a big data approach, they applied machine learning to create classifiers able to identify the presence of cancer-derived DNA within [blood samples](#) and to determine what [cancer type](#). This basically turns the 'one needle in the haystack' problem into a more solvable 'thousands of needles in the haystack', where the computer just needs to find a few needles to define which haystack has needles.

The scientists tracked the cancer origin and type by comparing 300 patient tumour samples from seven disease sites (lung, pancreatic, colorectal, breast, leukemia, bladder and kidney) and samples from healthy donors with the analysis of cell-free DNA circulating in the blood plasma. In every sample, the "floating" plasma DNA matched the tumour DNA. The team has since expanded the research and has now profiled and successfully matched more than 700 tumour and blood samples from more cancer types.

Beyond the lab, next steps to further validate this approach include

analysing data from large population health research studies already under way in several countries, where blood samples were collected months to years before cancer diagnosis. Then the approach will need to be ultimately validated in prospective studies for cancer screening.

More information: Shu Yi Shen et al. Sensitive tumour detection and classification using plasma cell-free DNA methylomes, *Nature* (2018).

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