'Liquid Biopsy': Blood test gives 'real-time' picture of cancer

31 May 2012

(Medical Xpress) -- A simple and cost-effective blood test could be used to monitor how a patient is responding to treatment and detect genetic faults in their cancer as they happen, according to a Cancer Research UK study.

The test accurately measures the levels of 'faulty' DNA fragments that are shed into the bloodstream by cancer cells as they die. By tracking these levels, scientists at Cancer Research UK's Cambridge Research Institute were able to detect genetic faults involved in tumour growth in blood samples taken from twenty women with ovarian cancer.

The researchers were also able to build a 'real-time' picture of how one woman's breast cancer was responding to treatment over more than a year.

As treatments become more targeted towards genetic mutations, this approach could mean that a patient is given certain treatments based on the results of a quick blood test, sparing them from an invasive biopsy.

Study author Dr Nitzan Rosenfeld, based at the Cancer Research UK Cambridge Research Institute, said: "This type of blood test has the potential to revolutionise the way we diagnose and treat cancer. The great advantage is that it can be used to identify cancer mutations without surgery or a biopsy, making it safer and cheaper."

The test also overcomes one of the main limitations of tumour biopsies, where a sample may only give a limited snapshot of the mutations that are present in cancer. It's also difficult to take samples from secondary cancers throughout the body, once the disease has spread.

Because DNA is shed from all tumours into the bloodstream, this test gives a fuller picture of the disease's progress.

This is the first time scientists have been able to screen entire genes in a blood test to identify mutations that have arisen in the cancer, and it could transform how cancers are monitored and treated in the future.

The research, published today in *Science Translational Medicine,* used DNA sequencing to look for nearly 20,000 possible mutations in 6 cancer-related genes and was able to identify rare DNA molecules containing cancer-specific genetic faults.

The researchers identified mutations in blood samples from twenty ovarian cancer patients treated at Addenbrooke's Hospital in Cambridge. They were also able to measure changes in the amounts of cancer DNA in the blood stream in several ovarian and breast cancer patients.

Dr James Brenton, a Cancer Research UK ovarian cancer clinician and study author, said: "Our technique is much more comprehensive and practical than others that have been used to measure DNA in the blood. More than two per cent of the DNA we found in plasma of advanced cancer patients came from the tumour. This tumour specific DNA offers us an opportunity to follow the disease in 'real-time' as it changes, helping us to respond and change the treatments we use against the disease."

In one of the ovarian samples studied, the test was able to detect a new genetic fault not found in the original biopsy. After further examination, they found that this mutation was present in a small minority of cells in one part of the tumour, but could not be identified in most areas tested.

The researchers believe that implementing the test could be done at a similar cost to others used to detect and monitor cancer and a single technician could carry out several hundred tests a week.

Dr Rosenfeld added: "We need to confirm its accuracy in more patients, and in additional cancer types, but this test could be adapted to look for mutations in different cancers and updated to include new genetic faults as research uncovers them."

Professor Peter Johnson, Cancer Research UK's chief clinician, said: "These initial results hold out the promise of a new way to monitor cancer and how it can change as patients undergo treatment. In the future we will be using many more treatments aimed at molecular changes in cancer cells, and this gives us a system that could allow us to respond and modify treatments as we see new gene changes occurring."

More information: Forshew, T et al, Non-invasive identification and monitoring of cancer mutations by targeted deep sequencing of plasma DNA *Science Translational Medicine* (2012) DOI: 10.1126/scitranslmed.3003726

Provided by Cancer Research UK