Groundbreaking computer program diagnoses cancer in two days

October 7 2015

In by far the majority of cancer cases, the doctor can quickly identify the source of the disease, for example cancer of the liver, lungs, etc. However, in about one in 20 cases, the doctor can confirm that the patient has cancer—but cannot find the source. These patients then face the prospect of a long wait with numerous diagnostic tests and attempts to locate the origin of the cancer before starting any treatment.

Now, researchers at DTU Systems Biology have combined genetics with computer science and created a new diagnostic technology based on advanced self-learning computer algorithms which—on the basis of a biopsy from a metastasis—can with 85 per cent certainty identify the source of the disease and thus target treatment and, ultimately, improve the prognosis for the patient.

Each year, about 35,000 people are diagnosed with cancer in Denmark, and many of them face the prospect of a long wait until the cancer has been diagnosed and its source located. However, even after very extensive tests, there will still be 2-3 per cent of patients where it has not been possible to find the origin of the cancer. In such cases, the patient will be treated with a cocktail of chemotherapy instead of a more appropriately targeted treatment, which could be more effective and gentler on the patient.

Fast and accurate results

The newly developed method, which researchers are calling TumorTracer, are based on analyses of DNA mutations in cancer tissue samples from patients with metastasized cancer, i.e. cancer which has spread.

The pattern of mutations is analysed in a computer program which has been trained to find possible primary tumour localizations. The method has been tested on many thousands of samples where the primary tumour was already identified, and it has proven extremely precise. The next step will be to test the method on patients with unknown primary tumours. In recent years, researchers have discovered several ways of using genome sequencing of tumours to predict whether an individual cancer patient will benefit from a specific type of medicine.

This is a very effective method, and it is becoming increasingly common to conduct such sequencing for cancer patients. Associate Professor Aron Eklund from DTU Systems Biology explains:

"We are very pleased that we can now use the same sequencing data together with our new algorithms to provide a much faster diagnosis for cancer cases that are difficult to diagnose, and to provide a useful diagnosis in cases which are currently impossible to diagnose. At the moment, it takes researchers two days to obtain a biopsy result, but we expect this time to be reduced as it becomes possible to do the sequencing increasingly faster. And it will be straightforward to integrate the method with the methods already being used by doctors."

Researchers expect that, in the long term, the method can also be used to identify the source of free cancer cells from a blood sample, and thus also as an effective and easy way of monitoring people who are at risk of developing cancer.

Read the scientific article TumorTracer: A method to identify the tissue

of origin from the somatic mutations of a tumour specimen in *BMC Medical Genomics*.

Tumor tracer

A diagnostic method for determining the primary site of the cancer. The method combines genetics and computer science, and can analyse a biopsy from a metastasis, and on this basis provide a number of possible scenarios for where the cancer may have developed and indicate the probability of it being correct. At the moment, analysing a biopsy takes two days.

Provided by Technical University of Denmark