DNA blood test detects lung cancer mutations

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Cancer DNA circulating in the bloodstream of lung cancer patients can provide doctors with vital mutation information that can help optimise treatment when tumour tissue is not available, an international group of researchers has reported at the European Lung Cancer Conference (ELCC) in Geneva, Switzerland.

The results have important implications for the use of cancer therapies that target specific cancer mutations, explains Dr Martin Reck from the Department of Thoracic Oncology at Lung Clinic Grosshansdorf, Germany, who presented the findings at the conference.

Testing for the presence of these mutations in the tumour itself is not always possible, however studies have suggested that DNA from the tumour that circulates in the bloodstream of patients may provide similar information.

The large international ASSESS study aimed to compare the ability of blood testing to detect EGFR mutations with the more standard method of testing the tumour itself.

"We were really asking a question on behalf of patients," Reck said: "Is there a valid test that can identify an EGFR mutation and give me the opportunity for superior treatment, even if my lung tumour is not accessible for bronchoscopy or CT-guided biopsy? And, are the results of this blood test in agreement with the results of the 'gold-standard' tissue test?"

Overall, the study included 1162 matched tissue and blood samples. Comparison of the outcomes of EGFR testing in the two techniques showed an 89% rate of agreement between the blood test and tissue test. Plasma testing identified about half of the patients with EGFR mutations, compared to tissue testing (a sensitivity of 46%).

The tests in this study were not performed in specially selected central labs, but in local labs that are used for daily clinical routine. "This is important, because it does reflect the clinical reality and not a 'virtual' trial reality," Reck said.

"The results mean that for patients who do not have accessible tumour tissue, plasma testing for EGFR mutation turns out to be an attractive option to offer these patients adequate targeted treatment," Reck added.

Commenting on the study, Dr Rafael Rosell, from the Catalan Institute of Oncology, Barcelona, Spain, expert on the ESMO Faculty on Lung cancer, said: "Cell-free DNA detected in the bloodstream of cancer patients represents an excellent tool to examine genetic alterations that are usually found through tumour tissue testing. This represents one of the most astonishing phenomena in biology."

"The results of this study validate that the presence of EGFR mutations in circulating DNA from plasma or serum (fractions obtained from whole blood) can be detected in around half of the patients."

Already, since this study was performed, improvement of techniques have seen the sensitivity of tests for EGFR mutations in circulating tumour DNA increase further, Rosell noted.

"This work paves the way for further studies and expands the routine use of examining mutations such as EGFR mutations as part of cancer patient care," Rosell said.

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