A universal cancer test is still a medical holy grail

The lifeblood coursing through your veins may one day become a messenger of death. Scientists revealed last week that they have taken baby steps towards developing a blood test for cancer. The so-called liquid biopsy looks for traces of mutated DNA and rogue proteins shed by tumours. For all the claims, it is not a universal test for this feared emperor of maladies. And, while earlier detection would be a boon to some, it could also become a burden for those whose modest abnormalities would not have proved life-threatening.

This new tool, being developed at Johns Hopkins University, is called CancerSEEK. The test looks for traces of 16 genes and eight proteins associated with eight types of cancer. The checklist included ovarian, liver, stomach and pancreatic cancers. All tend to be diagnosed late and have low survival rates.

Blood samples were taken from 3,000 cancer patients whose tumours had not spread. CancerSEEK flagged up about 70 per cent as showing evidence of the disease (a sensitivity of 70 per cent). This means that 30 per cent of cases were missed. They represent the false negatives: instances in which the disease exists but the test fails to expose it. The test's sensitivity fell to 43 per cent when it was trialed on the earliest cancers (so-called stage I tumours).

CancerSEEK fared much better when it came to ruling out who did not have cancer: of more than 800 healthy controls, only seven tested positive (false positives). The trouble is that controlled trials contain just two categories of people: the diseased and the completely healthy. Many of those likely to undergo a cancer test will have other common illnesses. Observers have noted that, since the test uses proteins that are also produced by diseases such as arthritis, the frequency of false positives may rise.

The Johns Hopkins research highlights the imprecise art of clinical screening and testing. Any test should identify the diseased without alarming the healthy; it should be cheap, safe and ideally non-invasive (CancerSEEK costs a respectable $360); and early detection should be accompanied by the realistic prospect of meaningful treatment. Usually, the earlier a cancer is picked up, the better; a blood test that can spot traces in asymptomatic individuals would be a game-changer. One company chasing this goal, launched in 2016 and whose backers include Amazon founder Jeff Bezos, has the revealing name of Grail.

Misdiagnosis, though, carries high costs for both patient and society: false positives can damage individuals psychologically (such as increasing suicide risk); dent public confidence; and incur expensive, unnecessary investigations. False negatives have an obvious cost in failing to catch treatable disease early.

Questioning the value of universal screening and testing does not mean advocating for ignorance. But it does mean considering the consequences of a positive test result.

Just such a debate has happened with prostate cancer. Although 16 per cent of American men will develop this cancer, only one out of six with a diagnosis will die from it. The prostate-specific antigen test used to screen for the disease cannot distinguish between harmless tumours and aggressive ones. Nonetheless, an elevated PSA result usually leads to further investigation and treatment — some of which affects mood and libido. And yet, for the majority of men, these serious interventions are for nothing. While one European study found some survival benefit among those taking the test, an equivalent American study did not. Today, men are advised to slip the PSA test unless they have particular concerns, such as a family history of the disease.

As one genetic counsellor told MIT Technology Review recently: “The big problem . . . will not be finding cancer, it will be finding the cancer that should be treated.”

The writer is a science commentator