HEALTH

# DNA test at local surgery will give warning of cancer and cardiac risks

British scientists have developed a way to screen patients for "actionable" mutations — faulty genes that increase the risk of diseases but can be mitigated through lifestyle or treatment

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Millions of adults will be offered a DNA test at GP surgeries to detect their risk of cancer and heart disease under plans for a "healthcare revolution".

A study has revealed that one in four people carry potentially harmful genetic mutations that can be picked up through a simple blood sample.

British scientists have developed a way to screen patients for "actionable" mutations — faulty genes that increase the risk of diseases but can be mitigated through lifestyle or treatment.

For example, patients with a gene linked to high cholesterol could be put on statins at a young age, while women with the BRCA breast cancer gene can be offered a preventative mastectomy.

The study leader, Professor Ros Eeles from the Institute of Cancer Research in London, said the test, costing £1,000 per patient, could be widely available at GP surgeries in two years.

Talks are being held with the NHS to make genetic screening a routine aspect of care that could save thousands of lives a year. This would make Britain a world leader in the use of whole- genome sequencing, a technique that maps out a person's entire DNA.

Over the past two years 102 healthy adults have taken part in the trial, which screened them for 566 genes linked to diseases, including heart conditions and blood-clotting disorders.

Overall one in four people had genes that cause disease, of which half were linked to cancer, such as the BRCA mutation carried by Angelina Jolie.

One woman who took part in the trial chose to have surgery to get her ovaries removed as a preventative measure after being told she had a gene linked to ovarian cancer. Four in ten had genetic changes linked to difficulties tolerating medicines such as antibiotics, meaning they can be given alternatives to reduce side effects.

Sajid Javid, the health secretary, said that the trial being presented at the American Society of Clinical Oncology conference in Chicago, demonstrated "genomics is changing the future".

He said: "This study shows the potential for genome sequencing in enabling patients with life-changing diseases to receive early diagnoses just by visiting their GPs."

The study also found that six in ten adults carry a recessive genetic alteration, which means they don't have the condition but could pass it on to their children.

Eeles, a consultant oncologist at the Royal Marsden NHS Foundation Trust, said that routine screening could prevent couples planning a family from passing on genetic diseases. Women who carry harmful mutations could undergo IVF and have embryos screened genetically, implanting those that did not have the mutation.

Eeles said it would take "two to five years" for the screening to be routinely available on the NHS. The next step is an "implementation study" involving 1,000 patients to see how the tool can be used at surgeries. "When we translate this into the NHS we want a system that's going to be as streamlined, fast and as cheap as possible," she said.

"We'd like to get [the cost per person] down to under £1,000. It sounds like a lot but if you prevent someone having a cancer . . . we're talking about £200,000 for two years of immunotherapy.

“It’s definitely coming and the UK is the leading area in the world to integrate whole-genome sequencing into a national health system.”

Patients could be offered the genomic screening when they register with a new GP and results could be stored on the NHS app. Many of the patients in the trial were put on new treatments after finding they had particular genes.

For example, some who had genes putting them at greater risk of cancers were being offered greater monitoring to spot the cancer early.

The study did not involve anyone younger than 25 to ensure the results did not cause distress.

Eeles added: “We don’t think there’s any point in looking for mutations that we can’t help people with. We’re in the business of doing things that can make a difference to people’s healthcare.”

Dr Michael Sandberg, a GP at 90 Sloane Street in London, which co-ordinated the research, said: “This study is pushing the boundaries of genomic screening by showing that it is feasible as part of GP care and has the potential to significantly improve people’s health.”

The NHS is increasingly using genetic data as part of patients’ treatment. A plan launched in 2019, said it wanted to be “the first national healthcare system to offer whole-genome sequencing as part of routine care”.

Unravelling a person’s genetic code to look for clues as to what was making them ill — or could put them at risk — was once the stuff of science fiction (Kat Lay writes).

Such medicine is now a routine part of NHS care. Cancer patients have samples taken from their tumours for genetic testing so doctors can choose the drugs best suited to target their condition’s weakness.

Whole genome sequencing is providing answers for people with rare diseases, who can find out the reasons for their symptoms. Even where a treatment does not exist, understanding the underpinnings of a disease means that scientists seeking a cure are pointed in the right direction.

The NHS Genomic Medicine Service, one of the first in the world, offers a suite of tests covering more than 3,000 rare diseases and over 200 types of cancer.

The screening programme fits with the NHS’s prevention agenda aimed at keeping people healthier for longer, rather than stepping in once they have become ill.

What is crucial is that the scientists behind these tests are only looking for "actionable" mutations — faulty genes that increase the risk of a disease but ones we can do something about.

Experts have long warned that telling someone they are at risk of a disease such as dementia — but that nothing can be done — would simply increase distress.

Ethical questions remain, however. Could a test showing an increased risk of heart disease also result in increased insurance premiums? And if lifestyle changes would mitigate the risk but the patient ignores them, should the NHS pay for treatment?