DNA testing 'revolution' at the GP could detect patients at risk of cancer

Using genomic sequencing in primary care means patients can get early treatment for health conditions before they become life-threatening

By Lizzie Roberts, HEALTH CORRESPONDENT, IN CHICAGO
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The introduction of genomic sequencing at GP surgeries could help a quarter of patients get early treatment for disease, the first UK study of its kind has found.

One in four people carry genetic mutations that could be specifically targeted with preventative treatments, so-called "actionable" genetic alterations, according to the research led by The Institute of Cancer Research (ICR) and The Royal Marsden NHS Foundation Trust.

Actionable genetic alterations increase the patients' risk of diseases like cancer, heart disease and blood clots.

Early modelling based on the study findings suggest thousands of lives a year could be saved by the tests, the researchers said, by reducing mortality by at least a fifth for breast and ovarian cancer patients.

The Health Secretary said on Thursday that the technology was "changing the future of healthcare", and opened up the possibility for patients with life-changing illnesses to be diagnosed early via their GP.

The process, known as whole genome sequencing (WGS), involves extracting patient DNA from a sample of blood, tissue or skin, and running it through a machine for analysis. Experts then examine the DNA for genetic mutations.

Sajid Javid has previously said babies will have their whole genome sequenced at birth to screen for genetic disorders.

But this new research, presented at the American Society for Clinical Oncology's annual conference, which begins on Friday in Chicago, has proven it is feasible to carry out the tests through primary care and would also benefit adults.

NHS rollout

The tests could be available on the NHS within two to five years' time, said Ros Eeles, professor of oncogenetics at the ICR and study lead, and could form part of an initial GP registration.

The team are in discussions with NHS England about a potential rollout for the tests, which cost around £1,800 per patient.

"It sounds like a lot, but ... to treat somebody with metastatic disease you could be talking about £200,000 for two years of immunotherapy treatment," Prof Eeles added.

TIMELINE

Timeline - Genetic breakthroughs

December 1999

First human chromosome is sequenced

April 2003

Heads of government of Britai

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Some 102 patients had their entire genetic code sequenced after samples were taken via a GP practice, and 26 had genetic mutations, according to the study.

Six in 10 patients also carried an inherited mutation in a "recessive gene", meaning they risked passing on the mutation to their children if their partner also carried it.

One person in the study was identified as having an increased risk of ovarian cancer and decided to have her ovaries removed preventatively.

Around 70 per cent of the patients with actionable mutations would not have been identified through standard medical registration and family history alone, the researchers said.

"Our study is the first to assess whether whole genome sequencing can be delivered by GPs and demonstrates that it is feasible, provides important genetic information, and is likely to benefit patients by making preventative measures or counselling available to them," Prof Eeles said.

"We feel that this work could place us at the start of a revolution for healthcare, by helping to pave the way for a future where genomic screening is provided routinely to patients in primary care."

'Genomics is changing the future of healthcare'

The patients were recruited from The London Genetics Centre, a private GP practice, between 2020 and 2022, and had their entire genetic code read from samples taken at the surgery.

Researchers looked for 566 genetic changes linked to disease, including 84 associated with cancer and 77 related to heart conditions. Detecting these mutations would change the way doctors manage patients' care, for example by offering increased monitoring and screening.

Some 38 participants also had genetic changes in their code which are linked to how patients respond to, or break down, certain medicines. Doctors can use this information to minimise potential side effects for patients from certain drugs, or help them decide to avoid particular treatments altogether.

Sajid Javid said: “Genomics is changing the future of healthcare, and this study shows the potential for genome sequencing in enabling patients with life-changing diseases to receive early diagnoses just by visiting their GPs in future.

“The UK continues to lead the way in genomics expertise, and this exciting collaboration between the Institute of Cancer Research, the NHS and the wider health sector has the potential to save lives by increasing the detection and prevention of diseases such as cancer and heart disease.”

The study also worked in partnership with cardiology consultants at The Royal Brompton and Harefield Hospitals.