MedicalXpress

Screening patients' genomes to allow GPs to assess disease risk is feasible

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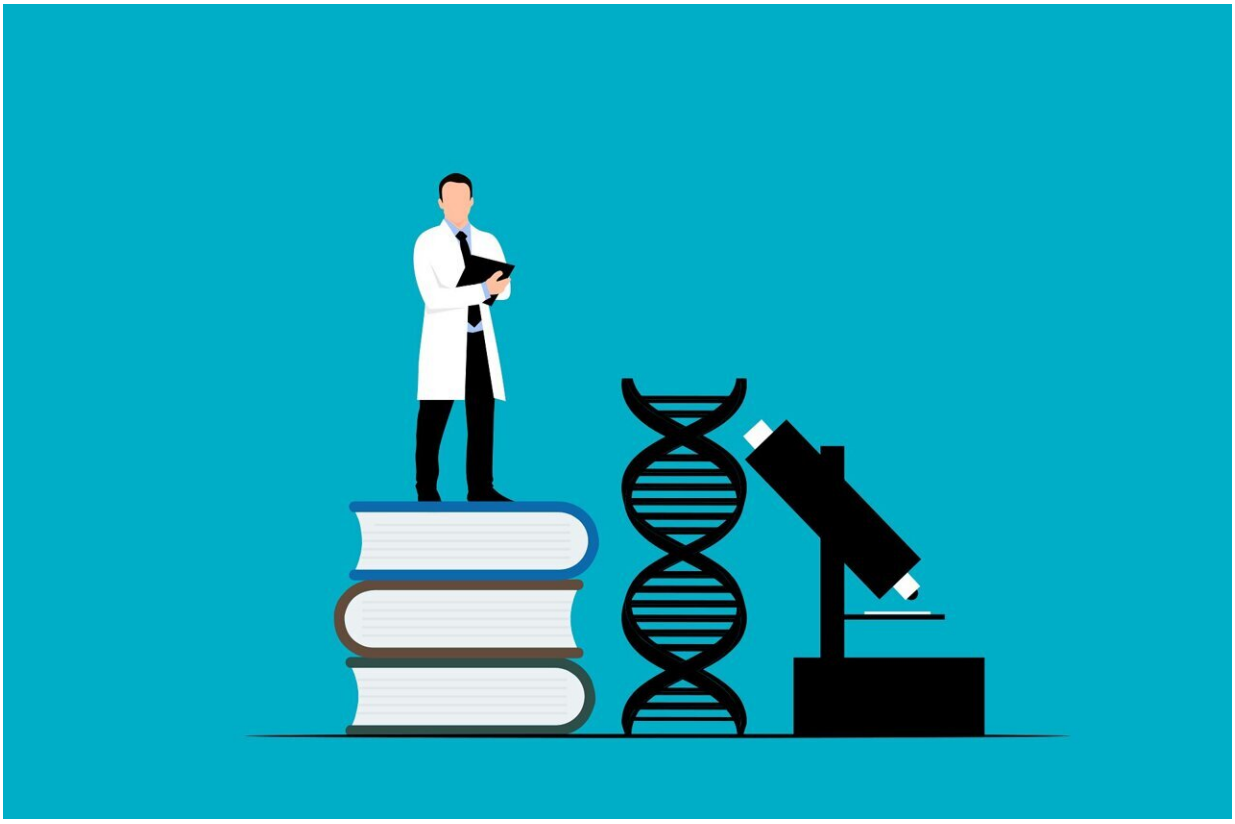

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Screening people's genomes as part of routine primary care can spot genetic changes that may have important implications for health, a new study reports.

The 90S study is the first to demonstrate that whole-genome sequencing (WGS) is feasible in a primary care setting and suggests it could have a role in detecting or preventing diseases like cancer or heart disease.

A quarter of people involved in the study had potentially "actionable" genetic alterations increasing their risk of diseases like cancer, heart diseases and blood clotting diseases.

The researchers also found that six in 10 people carried an inherited mutation in a "recessive" gene, meaning they wouldn't display any resulting condition themselves, but might risk passing it on to the next generation if their partner also had the same recessive genetic alteration.

Setting up a model pathway in primary care

A team at The Institute of Cancer Research, London, and The Royal Marsden NHS Foundation Trust, worked in partnership with cardiology Consultants at The Royal Brompton and Harefield Hospitals and with The London Genetics Centre at 90 Sloane Street, a private GP practice, to carry out the study.

The study was primarily focused on setting up a model pathway in primary care and showing its feasibility. The researchers stressed that they will need to do more work to demonstrate the benefits for patients of having alterations detected. They also want to work with the NHS to exploit these results and explore how to incorporate genetic data into general practice health screening more widely.

Results presented at ASCO annual meeting

The researchers will be presenting findings from the study at the American Society of Clinical Oncology annual meeting in Chicago.

The study involved 102 healthy participants, recruited from The London Genetics Centre at 90 Sloane Street between 2020 and 2022, who had their entire genetic code read from samples taken at the practice.

Researchers looked for 566 separate genetic changes linked to disease, including changes to 84 genes related to cancer and 77 related to heart conditions, and other genetic changes linked to how patients respond to or break down certain medicines.

Some 26 out of the 102 participants had potentially actionable genetic variants, 61 had a recessive gene that could be passed down to children should their partner also carry it, and 38 had genetic changes linked to specific responses to medicines.

Looking for 'actionable' genetic changes

The researchers said that detecting "actionable" genetic changes could trigger a change in the way patients and their families were managed by their doctor—either by offering monitoring, screening, or other measures to prevent disease.

Detecting recessive mutations that could cause disease in future generations would not affect the person's own health directly, but could have some benefits in offering support and genetic counseling to those who plan a future pregnancy, for example.

Information on how people break down drugs could help doctors minimize potential side effects for patients or help them decide whether they should avoid a particular treatment completely.

The 90S study is looking for gene alterations which if detected would alter choices for an individual such as lifestyle improvements, specific screening and sometimes targeted treatments. It will not report on the

risk of diseases for which there are no current actions that can be taken.

Evaluating risks and simplifying processes

Researchers will also be evaluating the risk of overdiagnosis—spotting at-risk cases that would not go on to cause disease.

The team aimed to make genetic screening practical and psychologically acceptable to participants by providing specially trained staff—a clinical fellow, research nurse, GPs and consultant geneticists, who could help them adjust to the medical and psychosocial consequences of any genetic findings.

The study leaders acknowledged that this level of resource may not be readily available on the NHS and they will be looking for ways of simplifying and improving processes to be suitable for large-volume NHS screening.

Study leader Professor Ros Eeles, Professor of Oncogenetics at The Institute of Cancer Research, London, and Consultant in Clinical Oncology and Oncogenetics at The Royal Marsden NHS Foundation Trust, 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 counseling available to them.

"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.

"Our findings suggest that integrating whole-genome sequencing into

primary care could change the way most patients and their families are managed by their GPs—either by detecting genetic variants that will affect a patient's own health, or by highlighting their risk of passing on a genetic alteration which is important for future family planning.

"However, the 90S study has shown us that this is not straightforward and so next, we want to start looking into how to best implement genomic screening as part of family practice within the NHS—seeking ways to simplify and improve processes and make it suitable for large-volume NHS screening."

Pushing the boundaries of genomic screening

Dr. Michael Sandberg, General Practitioner at 90 Sloane Street and Co-Principal Investigator for the 90S Study, 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. A large team of geneticists and specialists worked with our general practice to develop this model. We hope we could also stop many genetic conditions from being passed on to future children and grandchildren, if they have a 'monogene' basis, such as BRCA gene alterations. Whole genome screening could therefore have a huge impact and enormous benefits for people—and it is important to note we are only testing for genetic changes where we can help, so called 'actionable.'"

Provided by Institute of Cancer Research