We must not rush to implement new genetic screening programs, experts warn

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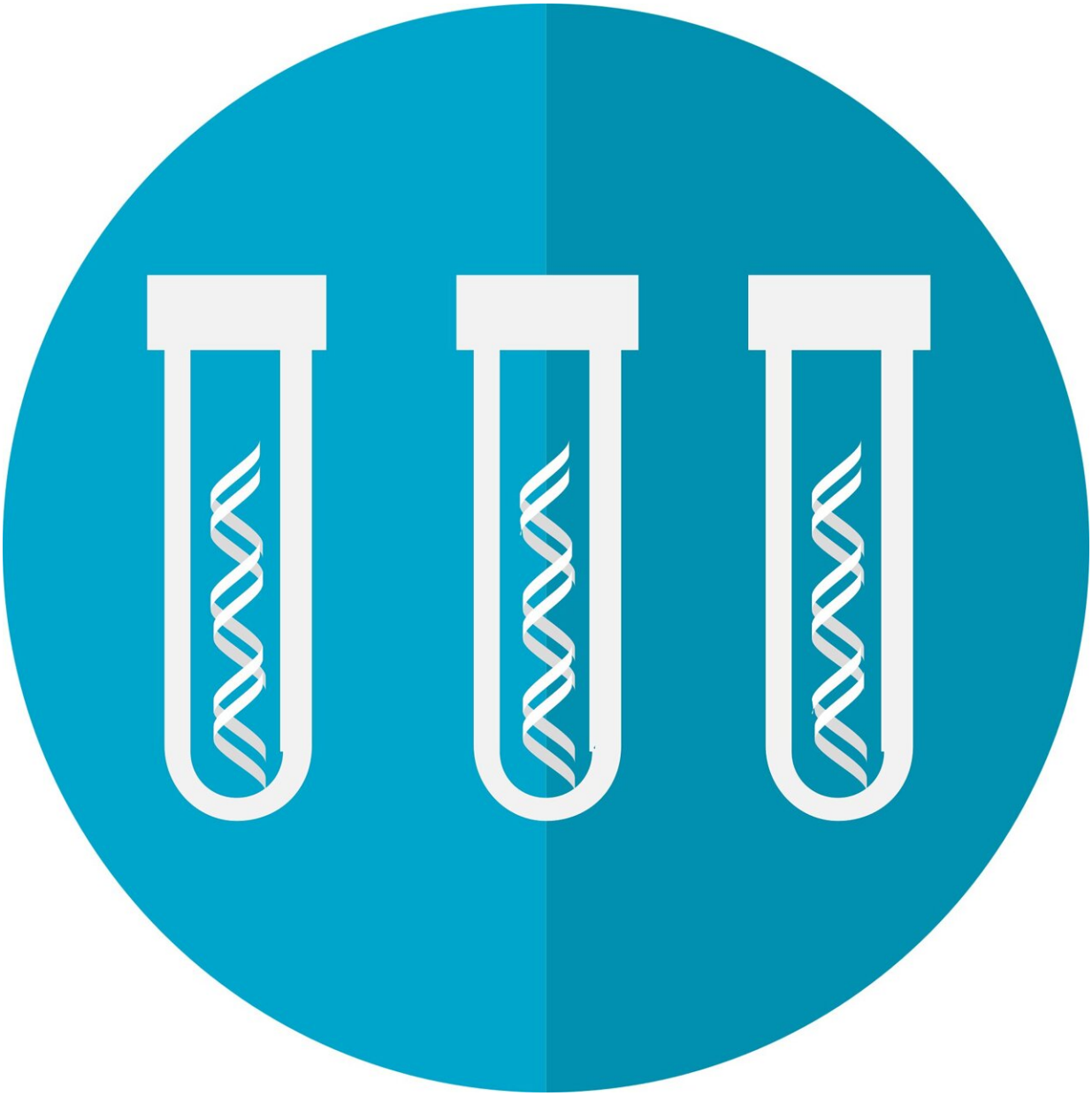

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There is a danger that population genetic screening programs may be introduced without the necessary scrutiny and rigor that is usually applied when screening programs are put into effect, experts have warned.

In an opinion piece published in *The Lancet*, a group of 12 leading U.K. geneticists, made up of top researchers and senior NHS clinicians, have expressed concerns that population screening using genetic technologies may be introduced without sufficient evidence that they are delivering net benefits for patients and the NHS.

The authors include leading academics from The Institute of Cancer Research, London, the University of Oxford, the University of Exeter, the University of Cambridge and The University of Manchester.

The authors highlight two major U.K. Government-backed genomic data research initiatives, the Newborn Genomes Program (also known as the Generation Study) and the Our Future Health program, both launched this year. In the Newborn Genomes Program, genetic data from whole genome sequencing for more than 200 diseases will be returned for 100,000 babies to their parents.

Participants in Our Future Health will receive results on genetic tests to predict whether they are at higher risk of common disorders such as cancer, diabetes and heart disease.

The group argue that, although valuable in providing data for researchers, these studies are not correctly designed to answer the key questions of any new screening program:

- Overall, will it make those participating live longer and better lives?
- Will the benefits from the screening outweigh the harms?

They highlight how new screening programs are normally subject to detailed scrutiny from experts in the National Screening Committee but that this important review process may be circumnavigated to expedite some of these higher profile new screening initiatives.

They suggest enthusiasm to apply genomic screening is being driven by commercial pressures, governmental early diagnosis targets, and a hopeful public, rather than robust evidence of benefits from these screening approaches.

Powerful tools

Genetic tests provide a powerful tool for rapid diagnosis of rare diseases once a patient has experienced symptoms, and for spotting people who carry well-understood mutations which strongly raise their risk of disease—such as BRCA1 and BRCA2 in breast and ovarian cancer.

But advances in the genomics field have sparked great enthusiasm in applying genetic testing on a population level, for example genetic risk scores that purport to rank people according to their disease risk. In their article, the experts warn that even for the most well-studied of diseases like breast cancer, genetic scores can in no way accurately predict who will, and who will not develop a disease.

Breast cancer, for example, has one of the best studied genetic risk scores, yet only 37% of breast cancer cases occur among people classified by the genetic score as being high-risk (in the top 20% of genetic risk scores). The majority of cases, 63%, occur in people who are not included in this genetic high-risk group.

This leaves people classified as low risk receiving false reassurance that they are unlikely to develop a disease, and many people classified as high risk are left anxious, even though they are overall unlikely to develop disease.

Professor Clare Turnbull, professor of translational cancer genetics at The Institute of Cancer Research, London, said, "The proposal of screening for a disease and catching it earlier is always attractive, and involving genetics might sound promising for improving screening. However, all screening programs cause some harms and utilize health care resources. We are concerned that these new genetic screening programs may be rushed through without making sure there is good evidence that they are having a positive effect on the key patient outcomes of survival and quality of life."

More information: Clare Turnbull et al, Population screening requires robust evidence—genomics is no exception, *The Lancet* (2023). DOI: 10.1016/S0140-6736(23)02295-X

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