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"Big Gene" industry in the NHS—does this represent good value?

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The ongoing political project to supposedly save the NHS continues. The past decade has mostly been filled with ideas that sound great, but don't have much evidence to back them up. We have had the era of artificial intelligence to triage patients, with claims it had greater accuracy than doctors[1]; management consultants—£640 million spent in three years alone[2]—and that's before we've got to the waste of marketisation, commissioning, and the National Programme for IT (dismantled in 2011 after costing billions).[3]

Now we have reached the "Big Gene" era. One example of this is the Our Future Health (OFH) project.[4] It's aiming to be "the UK's largest ever health research programme," "designed to help people live healthier lives for longer through the discovery and testing of more effective approaches to prevention, earlier detection, and treatment of diseases." It is predominantly funded by the life sciences industry, but also by the government and charities. The project expects to recruit around five million patients from primary care who will have their blood taken and DNA extracted. Their polygenic risk score will be calculated, and they will then be invited to join further studies. These will mainly be run by industry, who will be able to have their own systems accredited to store de-identified data from participants.

We need high quality, clinical trials in medicine, but we also need to invest in projects wisely. Can polygenic risk scores realistically be expected to meet the aims of the project? This is screening—a complex intervention where a test is one small part of a larger process. Polygenic risk scores rank people at higher or lower-than-middling risk, but one must recall basic statistics on predictive values. I will save you the effort—the bigger the attempt not to miss people with serious disease, the more people have to be screened; the more people are screened, the more false positives. Restrict the test to people at high risk, and the volume of false positives will decline, but since diseases usually falls in the larger group of lower risk people, you will also mainly miss it. This is why effective disease screening is exceptional: it rarely works well enough to do more good than harm.

Then there is the Galleri trial, where participants' blood is tested for abnormal methylation of DNA, in what is described as a "multi-cancer early detection test." However, what happens next to this information is similar: again, it's about risk stratification, with participants referred on for imaging or other investigations for "cancer signals."

When false positives are so inherent, it's not just about the actual money spent on the contract, or the potentially false promises of overhyped projects, but also the time required to deal with any fallout in the NHS. Providing the scans and having discussions about test results will take huge amounts of time for staff —competing with people who are symptomatic and languishing on waiting lists. Although these projects are substantially funded by industry, it will be the NHS that's left to pick up the pieces—does this really represent good value for the NHS?

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2 Oliver D. Stop wasting taxpayers' money on management consultancy for the NHS BMJ 2014; 349:g7243 doi: 10.1136/bmj.g7243
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4 Our Future Health. https://ourfuturehealth.org.uk/about-us/