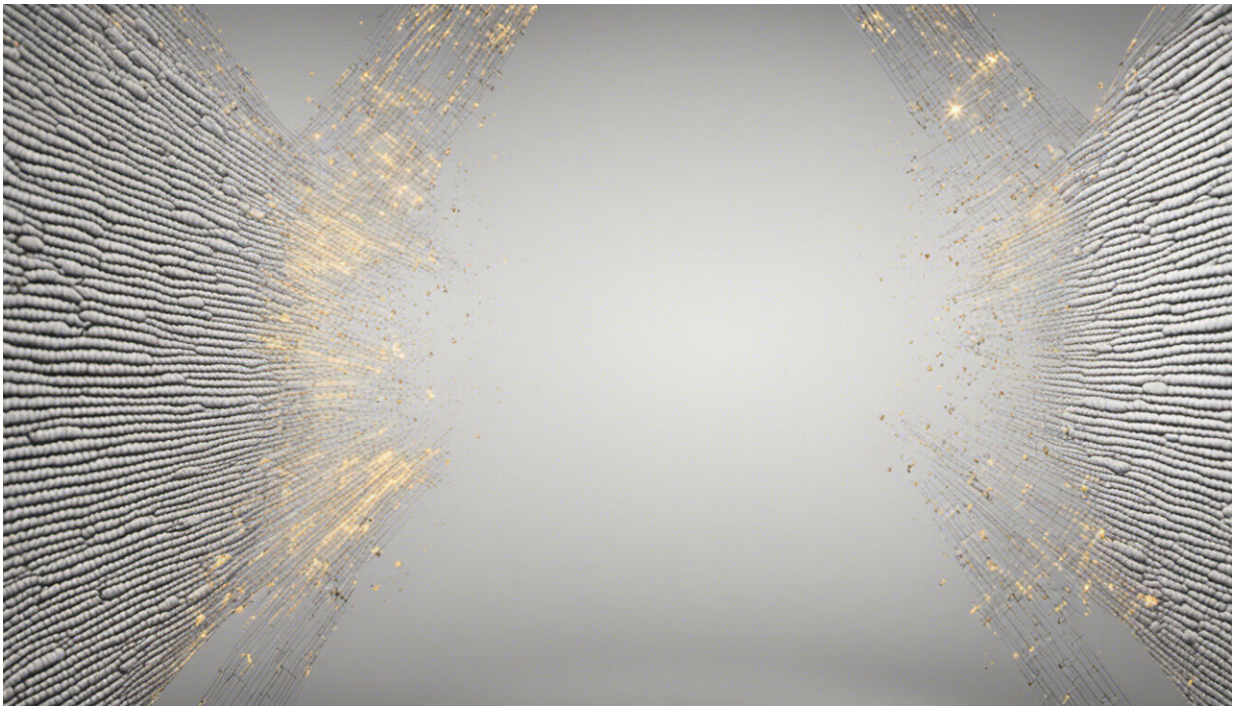
# Opinion: Why we should be worried about gene-carrier screening

August 2 2017, by Felicity Boardman



Credit: AI-generated image (disclaimer)

The ability to cheaply and quickly sequence entire genomes is changing the way diseases are identified and treated. But it is also likely to change the way we make some of the most important and personal decisions of our lives: how, and with whom, we have children.

Pre-conception genetic screening (testing for "carrier status" before pregnancy) has usually only been available to couples already known to be at risk of a particular disease. Ashkenazi Jews, for example, are most likely to be carriers of the mutated HEXA gene linked to Tay-Sachs disease, a fatal genetic disorder. Screening for the HEXA mutation is therefore recommended for all Ashkenazi Jews planning children. Today, though, genome sequencing is more affordable and accessible than ever before. It is now (at least, technically) possible to screen everyone to find out if they are a carrier of a genetic disease.

These technological changes have triggered increased enthusiasm for carrier screening. Recently, the American College of Obstetricians and Gynaecologists recommended that carrier screening be offered to everyone, and at the very least to all pregnant women. Similarly, private companies in the UK are now offering pre-conception screening for large panels of genetic conditions, simultaneously.

It is estimated that the average person carries three to five serious genetic conditions, such as spinal muscular atrophy or cystic fibrosis, so the routine use of pre-conception screening could affect a lot of people and the way they approach relationships.

The "genetic compatability" of a couple is set to become an important consideration for future generations, changing the way relationships are formed. Dating apps, for example, which give users snapshot information about a potential mate, could become places where carrier status is disclosed alongside other notable physical characteristics such as hair and eye colour.

Some of these apps are already emerging – one has been dubbed Tinder for Tay-Sachs. Indeed, through the Dor Yeshorim initiative, the Ashkenazi community has long operated an anonymous programme of pre-marital genetic screening for this very purpose.

Unlike an Ashkenazi Jew, who may be more prepared for the news that they are a carrier of Tay-Sachs, research shows that most people who undergo carrier screening are entirely unprepared for a positive result. As most genetic disorders screened for are considered rare, those having the screening will probably have never experienced, or even heard of, the condition they have been found to carry.

## Impossible judgement

This lack of experience is significant because research shows that it makes an important difference to the way people make decisions about having a baby with that condition. For example, a recent study my colleagues and I conducted showed that people without prior experience of spinal muscular atrophy took a much dimmer view of the condition than families who live with it. They were also more likely than families with spinal muscular atrophy families to consider termination of an affected pregnancy as acceptable.

This contrast highlights that decisions about whether to introduce pre-conception screening, and which conditions it should be used for, are not as straightforward as they initially appear. What may be a liveable disability to one person, may be an intolerable experience to another. Yet pre-conception genetic screening calls on us to make this impossible judgement before that person is even conceived.

Given the wide variability and unpredictability of genetic disease, it may appear logical to avoid all forms of it – as far as current technology allows, anyway. However, the emergence of groups, such as "don't screen us out", which lobby against screening for Down's syndrome, highlight the difficulties that surround such blanket avoidance.

Many people can, and do, live happy and fulfilling lives with a genetic disease. Yet, once introduced on a mass scale, screening programmes

can become difficult to refuse. Having a disabled child, when the technology existed to prevent it, could come to be seen as knowingly inflicting that condition on the child.

While personal choice and control are at the heart of the drive towards pre-conception screening, we also need to consider what other choices become closed off by its introduction. Neutralising the genetic gamble does not come without a cost. And, as a society, we need to carefully consider just what sacrifices we are prepared to make for it.

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