

# NUFFIELD COUNCIL ON BIOETHICS

## Submission to the House of Commons Science and Technology Committee inquiry on commercial genomics

26 April 2019

### Key points

- Developments in commercial genomics can provide tools to enable people to take more responsibility for their health. However, there are potential harms as well. There can be: consequences for individuals having testing, for example if results are inaccurate or hard to interpret and/or poorly supported by robust research; social harms when private testing undermines equal access to healthcare and creates burdens on the NHS; and harms to third parties, for example if children are tested without consent.
- It is important that people receive their results in appropriate ways, are helped to interpret their results, and receive psychosocial support if necessary. Many genomic testing companies do not provide these services.
- It is not clear whether genomic testing companies' data storage, sharing and access policies meet important ethical principles. It would be helpful if the Science and Technology Committee would clarify current practice.
- We have particular concerns about the way non-invasive prenatal testing (NIPT) is offered in the private sector. Although we are pleased this issue is now gaining some attention, we have seen little actual change in practice.
- Overall, our impression is that regulation of the commercial genomics sector is light. The Committee might consider commercial genomic testing of children as an area where strengthened regulation or oversight needs to be introduced.
- The proposal to offer paid-for genomic testing to healthy individuals in the NHS raises a number of questions about purpose, clinical validity and ethical acceptability.

### Introduction

The Nuffield Council on Bioethics is an independent body that examines and reports on ethical issues in biology and medicine. This submission is drawn from the principles and conclusions set out in several of our publications, including:

- Bioethics briefing note: [Whole genome sequencing of babies](#) (2018)
- Report: [Non-invasive prenatal testing: ethical issues](#) (2017)

- Report: [The collection, linking and use of data in biomedical research and health care: ethical issues](#) (2015)
- Report: [Human bodies: donation for medicine and research](#) (2011)
- Report: [Medical profiling and online medicine: the ethics of 'personalised healthcare' in a consumer age](#) (2010).

We welcome the Science and Technology Committee's inquiry on commercial genomics. There has been considerable attention by policy makers on the utilisation of genomic technology in the NHS, but the commercial genomics sector has received less scrutiny.

It is difficult to obtain exact figures, but indications suggest the direct-to-consumer (DTC) genomic testing market is expanding rapidly across the world.<sup>1</sup> It is now possible to purchase a range of tests, including:

- Ancestry check (from around £80) – to find out information about family tree and relatives, including paternity.
- Health check (from around £100) – find out about a person's risk of developing a wide range of conditions such as breast cancer, Alzheimer's and Parkinson's disease.
- Telomere check (from around £70) – to track a person's 'cellular age'.
- Carrier screening (from around £100) – in anticipation of starting a family, to find out whether a person is a carrier of a recessive gene variant associated with genetics conditions such as cystic fibrosis.
- Prenatal screening (from around £375) – during pregnancy, find out the chance a fetus has a genetic condition such as Down's syndrome.
- Newborn screening (from around £300) – find out about whether your baby has a gene variant that is associated with a genetic condition.
- Whole genome sequencing (from around £800) – to acquire a person's full DNA sequence and store it with the company as a 'resource for life'.

## Ethical values

In the Nuffield Council's 2010 report on medical profiling, we identified five ethical values as being important when considering developments in medical profiling and online medicine, such as DTC genomic testing.<sup>2</sup> These are:

- 1 Private information should be safeguarded.
- 2 Individuals should be able to pursue their own interests in their own way.
- 3 The state should act to reduce harm.
- 4 Public resources should be used fairly and efficiently.

<sup>1</sup> Credence Research (2018) Press release: *DTC genetic testing market poised to grow with a double digit CAGR during the forecast period* <https://www.credenceresearch.com/press/global-direct-to-consumer-genetic-testing-market>

<sup>2</sup> Nuffield Council on Bioethics (2010) *Medical profiling and online medicine: the ethics of 'personalised healthcare' in a consumer age*, available at: <http://nuffieldbioethics.org/project/personalised-healthcare-0>

- 5 Social solidarity – sharing risks and working together to protect the vulnerable – should inform public policy.

These ethical values often conflict with one another. However, all are important and no one value invariably trumps another. In our report, we attempt to ‘soften’ the conflicts between these ethical values by seeking to align them as much as possible and making recommendations that are evidence-based, proportionate and feasible. If there is broad agreement that a development in DTC genomic testing is likely to cause significant harm, then intervention by governments or other organisations is justified.

Comments on the specific issues being explored by the Committee are provided below.

## **1 Any health or other benefits that consumers can derive from using commercially available genomic testing**

Policy makers in the UK and elsewhere often encourage people to take more responsibility for their own health, to lead a healthy lifestyle and play an active role in managing their healthcare. Developments in commercial genomics can provide tools to enable people to take more responsibility for their health. Genomic testing has the potential to allow early intervention, give more personal control of one’s health, save public healthcare resources if testing and treatment is conducted privately, and alert relatives to important genetic conditions or predispositions.

However, there are potential costs and harms associated with commercial genomic testing as well. These include:

- consequences for individuals if: a) results are inaccurate or hard to interpret and/or poorly supported by robust research; b) nothing can be done; c) unnecessary follow-up testing and treatment is carried out; d) inaccurate risk assessments lead to false reassurance or misplaced anxiety; and d) results lead to stigma or information abuse or other effects that may be regretted.
- social harms when private testing undermines equal access to healthcare and creates unnecessary additional burdens on the NHS;
- costs and harms to third parties, for example if children or third parties are tested without consent; and
- changes in perceptions of wellness and illness through medicalisation of normal variation.

### *Case study: non-invasive prenatal testing*

Genomic tests vary in the type and quality of information they provide, and how they are offered to people can affect the potential for benefit and harm. They should not all be treated the same. We considered the ethical issues raised specifically by the offer of non-invasive prenatal testing (NIPT) in different contexts in 2017.<sup>3</sup>

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<sup>3</sup> Nuffield Council on Bioethics (2017) *Non-invasive prenatal testing: ethical issues*, available at: <http://nuffieldbioethics.org/project/non-invasive-prenatal-testing>

NIPT can enhance reproductive autonomy by, for example, enabling women and couples to prepare for a baby with a genetic condition or impairment, or helping them decide whether to have a termination. NIPT is not yet available in the NHS across the whole of the UK and, when it is available, it will be offered only to women who have been found (through early pregnancy screening tests, such as the combined test) to have a higher chance of having a fetus with Down's syndrome, Edwards' syndrome or Patau's syndrome. Accessing NIPT for these conditions through the commercial sector provides a way for other women and couples, who have the financial means and who wish to, to find out important information about their fetus at an early stage of pregnancy.

We make a number of recommendations relating to how NIPT should be offered to women and couples:

- NIPT should only be offered for significant medical conditions or impairments that manifest in childhood. NIPT normally should not be used to test whether a fetus has a less significant medical condition, or an adult onset condition; to find out whether the fetus is the carrier of a recessive gene variant for any kind of medical condition; nor to reveal non-medical traits of the fetus (such as sex).
- NIPT should only be available within an environment that enables women and couples to make autonomous, informed choices, and when steps are taken to minimise the potential harms of offering NIPT. This, we suggest, involves the following:
  - NIPT should only be offered if it provides an accurate prediction of whether the fetus has or does not have the condition being tested for;
  - all providers of NIPT have a responsibility to provide high quality information and support to women and couples about the test and the condition being tested for; and
  - the Government should ensure it is meeting its duties to provide disabled people with high quality specialist health and social care, and to tackle the discrimination, exclusion and negative societal attitudes experienced by disabled people.

In our report, we highlight examples of how some commercial NIPT providers are not meeting these criteria, including:

- the misleading use of statistics and language about the accuracy of NIPT;
- poor or absent information about the tested-for conditions;
- offering NIPT for conditions (such as sex chromosome and microdeletion conditions) where the accuracy of testing is low or unknown;
- lack of follow-up support, such as access to a healthcare professional to discuss high chance results.

These failings have the potential to cause harm by creating confusion and anxiety among women and couples seeking NIPT services in the private sector, and to affect their ability to make informed choices.

Since we published our report, we have been urging companies, and relevant authorities, to improve the way that NIPT is offered to women and couples through the private sector. Although we are pleased this issue is now gaining some attention, we have seen little actual change in practice.

## **2 The extent to which currently available genomic sequencing and interpretation can provide accurate and unambiguous health results, for healthy and ill sections of the population**

We have an increasing understanding of the human genome and what it means for our health. UK projects such as the 100,000 Genomes Project are leading the way internationally to identify gene variants associated with rare diseases and cancers. However, the relevance of large amounts of the genome, and how different gene variants interact with each other, still remains largely unknown. Also, even if the effect of a gene variant in ill people or those with a family history is well understood, there is often little knowledge of what these variants mean for apparently healthy people. Clinical geneticists are discovering that some gene variants identified in symptomatic patients may have very different implications when those exact same variants are found in members of the general population.<sup>4</sup>

Despite this, several genetic testing companies offer genetic health risk tests for conditions such as Parkinson disease, late-onset Alzheimer disease, and hereditary hemochromatosis. This is based on the presence or absence of certain genetic variants in the sample. The tests often do not include all of the genes that have been associated with these conditions.

Some companies offer to provide people with raw sequencing data alongside their interpretation of the results, and people can go to other companies for further interpretation services. A study conducted in the US has highlighted that the misinterpretation and potential inaccuracy of raw data pose risks to individuals. Of 49 patients referred for clinical diagnostic testing with variants previously identified in the raw data from DTC genetic testing, the authors found that 40 per cent were false positives. Most of the variants tested were those associated with cancer. The authors conclude that information obtained from requesting and interpreting raw data could lead to inappropriate changes in people's care.<sup>5</sup>

## **3 The counselling or other support offered for those receiving, or considering asking for, commercial genomic test results, and whether this is to the standard required; the potential benefits and risks for the NHS that arise from the increasing availability of commercial genomic testing**

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<sup>4</sup> For example, see letter to Matt Hancock, Secretary of State for Health and Social Care, January 2019, from the Joint Committee on Genomics in Medicine of the Royal College of Physicians, Royal College of Pathologists, the British Society for Genetic Medicine (BSGM), and Clinical Genetics Society.

<sup>5</sup> Tandy-Connor S *et al.* (2018) False-positive results released by direct-to-consumer genetic tests highlight the importance of clinical confirmation testing for appropriate patient care *Genet Med* **20**:1515-1521.

The results of genomic tests can be shocking, distressing, uncertain and confusing. It is important that people receive their results in appropriate ways, are helped to interpret their results, and receive psychosocial support if necessary. Sometimes results might have implications for family members. In the NHS, this would be discussed before testing, and relatives could be contacted by healthcare professionals with the person's consent.

#### *Tests accessed through a clinic or hospital*

NIPT is usually offered through a private hospital or clinic. Some offer a package of care that includes pre-test counselling, access to a healthcare professional to discuss high chance results, and follow-up diagnostic testing if requested. But many do not offer all this. We have heard that women with high chance NIPT results are going to the NHS for advice, support and diagnostic testing. How the NHS will meet demand for this as private provision escalates is a source of concern.<sup>6</sup> Although maternity care staff are receiving training in preparation for the roll-out of NIPT for Down's, Edwards' and Patau's syndromes, they are less likely to know about the other conditions or results that private NIPT providers offer to test for.

Hence, we are pleased that the Care Quality Commission (CQC) has taken up our recommendation to inspect private clinics that offer NIPT. NIPT was previously considered to be exempt from the remit of CQC. In February 2019, Ted Baker, Chief Inspector of Hospitals at the CQC, said: "We expect providers of NIPT to ensure that women fully understand the procedure, know that it is not a diagnostic test, are informed about the possible outcomes, and that appropriate support is made available when delivering the test results. This includes facilitating access to counselling and other relevant services as well as medical follow up where this is needed." We hope to work with the CQC to help them determine exactly what a high quality NIPT service looks like.<sup>7</sup>

#### *Tests accessed through the internet*

Most commercial genomic testing can be accessed directly from companies' websites, without going to a clinic and having a face-to-face consultation with a healthcare professional. Counselling or other support for those receiving, or considering asking for, commercial genomic test results is not a standard requirement. Genetic specialists working in the NHS have told us that they are seeing an increase in the number of people being referred who have a result from a paid-for genetic test, particularly over the past year. NHS Genetics Centres have been advised not to see people if the test is not one that would be normally offered in the NHS (and therefore is of proven clinical validity and utility), but clinicians have told us that it can be difficult to turn people away. The NHS is already undergoing major financial stress, so this raises issues about the fair and efficient use of public resources.

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<sup>6</sup> See paragraph 4.35-36 of the Nuffield Council on Bioethics' [report on NIPT](#).

<sup>7</sup> Nuffield Council on Bioethics news story (8 February 2019) *Care Quality Commission to regulate private providers of non-invasive prenatal testing following Nuffield Council recommendation*, available at: <http://nuffieldbioethics.org/news/2019/care-quality-commission-regulate-private-providers-noninvasive-prenatal>

#### **4 What data obtained from genomic testing could be used for and if sufficient protection is in place for consumers using commercial genomic tests**

We considered the ethical issues raised by collection, linking and use of biological and health data in a report published in 2015.<sup>8</sup> Here we outline our core conclusions.

Individuals generally see controlling access to personal information, especially that relating to their health, as an important aspect of maintaining their privacy. If information is accessed or disclosed against their wishes, it can affect individuals' well-being and infringe their rights. Respecting people's privacy shows respect for them as individuals. However, there are circumstances in which it may be acceptable to challenge normal expectations of privacy.

We suggest that an ethically appropriate use of data should respect core moral standards, reflecting the basic rights that underpin the legal system, rather than simply aiming to satisfy the requirements of the law. A proposed course of action (i.e. in sharing of data) can be lawful, but still morally questionable. Involving people in the design and governance of a data initiative allows their interests and values to be expressed, transformed and reconciled. It can also help to secure their commitment to the outcome and build trust.

Our report sets out four ethical principles for the use of data in biomedical research and health care:

- The set of expectations about how data will be used in a data initiative should be grounded in the principle of respect for persons. This includes recognition of a person's profound moral interest in controlling others' access to and disclosure of information relating to them held in circumstances they regard as confidential.
- The set of expectations about how data will be used in a data initiative should be determined with regard to established human rights. This will include limitations on the power of states and others to interfere with the privacy of individual citizens in the public interest (including to protect the interests of others).
- The set of expectations about how data will be used (or re-used) in a data initiative, and the appropriate measures and procedures for ensuring that those expectations are met, should be determined with the participation of people with morally relevant interests.
- A data initiative should be subject to effective systems of governance and accountability that are themselves morally justified. This should include both structures of accountability that invoke legitimate judicial and political authority, and social accountability arising from engagement of people in a society.

It is not clear whether genomic testing companies' data storage, sharing and access policies meet our ethical principles, and it would be helpful if the Science and Technology Committee would investigate and clarify current practice.

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<sup>8</sup> Nuffield Council on Bioethics (2015) *The collection, linking and use of data in biomedical research and health care: ethical issues*, available at: <http://nuffieldbioethics.org/project/biological-health-data>

Particular areas that warrant attention include companies asking people for permission to use their genetic data for broad purposes such as 'research and product development', the potential for genetic databases to be target by hackers, and use of genealogy websites to discover previously unknown family relationships and the fact of donor conception.

There have been some well-publicised uses of commercial genomic databases for non-health purposes. In 2018, law enforcers in the US used the site GEDmatch to search for genetic data similar to that found at crime scenes of the notorious murderer and rapist known as the Golden State Killer. By finding relatives on the database with similar genomic profiles, they were then able to use genealogical information to identify a suspect, and then collect his DNA from a discarded cup.<sup>9</sup>

The Committee should be aware of the potential of blockchain technology to provide individuals with the ability to make informed choices about their genetic data. Blockchain-enabled platforms, it is suggested, could give individuals the ability to securely share their DNA data for specific purposes, such as a research project, either in order to help a cause that they believed in or for their own financial gain.<sup>10</sup>

## **5 The regulations or standards that commercial genomic tests are currently subject to, and if any new or strengthened regulations or standards should be introduced to mitigate any perceived risks associated with commercial genomic testing**

The manufacture of commercially-available genomic tests, and how they are marketed and offered to people, are regulated by different instruments and bodies. Overall, our impression is that regulation of the sector is light, and the international nature of the market raises challenges for policy makers in the UK.

### *Regulation of medical devices*

The manufacture of genomic tests in the UK is regulated by the Medical Devices Regulations, which implement the EU In-Vitro Diagnostic Medical Devices (IVD) Directive. This Directive has been replaced by the EU IVD Regulation, which will have direct force in the UK. Under the new Regulation, IVD manufacturers will be required to produce significantly more evidence on clinical performance of their tests. It has been suggested that this would be a step towards manufacturers becoming more responsible for the clinical utility of their devices.<sup>11</sup> The Regulation came into force in 2017, when it started a 5-year transition period. It will fully apply in 2022.

After the UK leaves the EU, it is likely that regulation of all medical devices will remain aligned with EU Regulation, though practical arrangements relating to certification and placing devices on the market are likely to change.<sup>12</sup> However, many manufacturers

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<sup>9</sup> See Scudder N *et al.* (2019) Policy and regulatory implications of the new frontier of forensic genomics: direct-to-consumer genetic data and genealogy records *Current Issues in Criminal Justice*, DOI: 10.1080/10345329.2018.1560588

<sup>10</sup> Schumacher A (2019) The era of monetised DNA? *Laboratory News*, available at: <https://www.labnews.co.uk/features/era-monetised-dna-21-02-2019/>

<sup>11</sup> BSI (2016) [How to prepare for and implement the upcoming IVDR – Dos and don'ts.](#)

<sup>12</sup> An overview of possible scenarios is provided by McHale J (2018) Health law, Brexit and medical devices: A question of legal regulation and patient safety *Medical law international* first published online.



of genomic tests are based outside Europe, where the IVD Directive/Regulation does not apply.<sup>13</sup> Moreover, the Directive only addresses the performance of the tests – not the ways in which it is offered or in which the service is provided.

### *Regulation of advertising*

Any advertising of products and services in the UK, including genomic testing, is monitored by the Committee of Advertising Practice (CAP), which produces codes of conduct for advertising in broadcast and non-broadcast media. The codes stipulate that advertisements must not be misleading, harmful or offensive. The rules in the codes are enforced by the Advertising Standards Authority (ASA).

In 2010, the Nuffield Council on Bioethics recommended that responsible authorities pay more attention to whether genetic test providers are making clinical claims for their products, even if implied rather than explicit.<sup>14</sup>

In 2017, we recommended that the Committee of Advertising Practice should more closely monitor the marketing activities of NIPT manufacturers and private hospitals and clinics to ensure that they are not misleading, harmful or offensive.<sup>15</sup> We are aware that the Advertising Standards Authority currently is in correspondence with several NIPT providers about potentially misleading content on their websites.

### *Regulation of services provided by clinics and hospitals*

Some commercial genomic tests usually can only be accessed through a clinic or hospital, such as NIPT. All health and social care providers in England, including private hospitals and clinics, must be registered and regulated by the Care Quality Commission (CQC) if they carry out one or more 'regulated activities' as described in the Health and Social Care Act 2008 (Regulated Activities) Regulations. The CQC inspects registered providers to assess how they are performing against fundamental standards of quality and safety set by Government, which are set out in the legislation. The taking or analysing of samples of tissue, cells or fluids in order to determine the existence of a genetically inherited disease or disorder, or to determine the influence of an individual's genetic variation on a drug response is exempt from the remit of the CQC (unless if carried out as part of the planning or delivery of the individual's treatment or care, or a national screening programme).<sup>16</sup>

In 2017, we recommended that the CQC should be inspecting clinics to ensure NIPT is provided to high standards of quality and safety. In 2018, the CQC reviewed its approach, taking into account our recommendations, and now include NIPT in the activities that are regulated by CQC.<sup>17</sup> However, the majority of commercial genomic

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<sup>13</sup> For a recent analysis of regulation of molecular diagnostic tests in the US, see Holloway K *et al.* (2019) Dangerous diagnostics? Regulatory reform in the genomic era *BMJ* 364:l640.

<sup>14</sup> Nuffield Council on Bioethics (2010) *Medical profiling and online medicine: the ethics of 'personalised healthcare' in a consumer age*, available at: <http://nuffieldbioethics.org/project/personalised-healthcare-0>

<sup>15</sup> Nuffield Council on Bioethics (2017) *Non-invasive prenatal testing: ethical issues*, available at: <http://nuffieldbioethics.org/project/non-invasive-prenatal-testing>

<sup>16</sup> Care Quality Commission (2015) [Registration under the Health and Social Care Act 2008. The scope of the registration](#)

<sup>17</sup> Nuffield Council on Bioethics news story (8 February 2019) *Care Quality Commission to regulate private providers of non-invasive prenatal testing following Nuffield Council recommendation*,

tests are carried out without the involvement of a clinic or hospital, and remain therefore outside the oversight of the CQC.

### *Testing of children*

Several companies offer, or are planning to offer, newborn screening tests that search for large numbers of genetic conditions. Parents of babies in the UK and elsewhere might be able to access whole genome and exome sequencing through commercial providers in the future. Whole genome sequencing is already available to adults through several US-based companies.<sup>18</sup>

Although not covered by any UK laws or regulations, there is professional guidance and advice on the use of direct-to-consumer genetic testing in children. There is general consensus within the medical genetics community that only information about childhood conditions should be shared with parents following whole genome sequencing of sick babies, and that it is not acceptable to use whole genome sequencing to look opportunistically for a broad range of conditions in babies. Examples of guidance include:

- Nuffield Council on Bioethics: “Companies should not carry out on children DNA tests that do not meet the criteria of the UK National Screening Committee.”<sup>19</sup>
- Human Genetics Commission: “Direct-to-consumer genetic tests in respect of children should normally be deferred until the attainment of capacity.”<sup>20</sup>
- European Society of Human Genetics: “Direct-to-consumer genetic tests should not be offered to individuals who have not reached the age of legal majority.”<sup>21</sup>

Concerns about genetic testing of healthy children often centre on the child’s right to an open future and their ability to make their own choices later about accessing their genetic information. Babies who have had their genome sequenced at birth might grow up to find that their genetic data has been stored in some format without their consent. Other concerns relate to how information obtained at birth could affect family expectations of the child, and how the child is raised.<sup>22</sup>

The Committee might consider DTC genomic testing of children as an area where strengthened regulation or oversight needs to be introduced.

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available at: <http://nuffieldbioethics.org/news/2019/care-quality-commission-regulate-private-providers-noninvasive-prenatal>

<sup>18</sup> See: [www.babygenes.net](http://www.babygenes.net) and [www.mybabygenome.com](http://www.mybabygenome.com).

<sup>19</sup> Nuffield Council on Bioethics (2010) *Medical profiling and online medicine: the ethics of 'personalised healthcare' in a consumer age*, available at: <http://nuffieldbioethics.org/project/personalised-healthcare-0>

<sup>20</sup> Human Genetics Commission (2010) A common framework of principles for direct-to-consumer genetic testing services.

<sup>21</sup> European Society of Human Genetics (2010) Statement of the ESHG on direct-to-consumer genetic testing for health-related purposes.

<sup>22</sup> Nuffield Council on Bioethics (2018) *Bioethics briefing note: Whole genome sequencing of babies*, available at: <http://nuffieldbioethics.org/project/briefing-notes/genome-sequencing-babies>

## **6 The potential benefits and risks, for individuals and for the NHS, and the ethical implications of the NHS offering genomic testing to healthy individuals willing to pay and share their data anonymously**

The proposal to offer paid-for genomic testing for healthy individuals through the NHS raises a number of questions about the purpose, clinical validity and ethical acceptability of such a scheme.

If the purpose was to gather genomic information for research, it would be uncommon, perhaps unheard-of, to ask participants to pay. Usually, participating in research does not incur financial costs for the participant, or they might be compensated in some way. We considered different kinds of transactions involved in the donation of bodily material for research in our report *Human bodies: donation for medicine and research* (2011).<sup>23</sup> We discuss the role of payment, but always in the context of the donor or volunteer *receiving* payment or reward of some kind in return for taking part in research. We also emphasise that altruism plays a central role in donation for research. An altruistic basis for donation helps underpin a communal and collective approach to the provision of bodily material for others' needs, where generosity and compassion are valued. Besides being an unusual way to recruit research participants, requiring people to pay would be likely to result in a skewed research population.

The scheme has more in common with a commercial transaction, where an individual receives goods or service in return for payment. The scheme would involve the genomic testing of healthy individuals, which suggests the person would receive information about their current and future health risks. Therefore, the purpose of the scheme might be to improve the health of those individuals taking part, by diagnosing disease or identifying risk factors for developing health conditions in future. This might, in turn, be expected to reduce the health burden on the NHS and save money. Offering genomic testing to healthy individuals through the NHS might also help mitigate some of the problems of the commercial provision of such tests, such as the lack of follow-up support and help in the interpretation of test results.

However, the clinical genetics community have raised concerns about the readiness of genomic testing to predict health outcomes in healthy people. For example, the Joint Committee on Genomics in Medicine of the Royal College of Physicians, Royal College of Pathologists, British Society for Genetic Medicine (BSGM), and Clinical Genetics Society stated in a letter to Matt Hancock, Secretary of State for Health and Social Care, in January 2019: "The potential for the NHS to save money by 'predictive' diagnosis has not yet been proven and it is more likely that variants of concern identified in a healthy individual would lead to NHS investigations and surveillance without clear evidence of benefit, with a net cost to the health care provider."<sup>24</sup>

If the purpose of the scheme was to improve health by diagnosing disease or identifying risk factors for developing health conditions in future, it could be considered

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<sup>23</sup> Nuffield Council on Bioethics (2011) *Human bodies: donation for medicine and research*, available at: <http://nuffieldbioethics.org/project/donation>

<sup>24</sup> Letter to Matt Hancock, Secretary of State for Health and Social Care, January 2019, from the Joint Committee on Genomics in Medicine of the Royal College of Physicians, Royal College of Pathologists, the British Society for Genetic Medicine (BSGM) and Clinical Genetics Society.

akin to a public health screening programme. There are strict criteria for the kinds of tests that can be offered within a screening programme. The UK National Screening Committee criteria for appraising the viability, effectiveness and appropriateness of screening programmes include:

Criteria 1. The condition should be an important health problem as judged by its frequency and/or severity.

Criteria 4. There should be a simple, safe, precise and validated screening test.

Criteria 9. There should be an effective intervention for patients identified through screening with evidence that intervention at a pre-symptomatic phase leads to better outcomes for the screened individual compared with usual care.

Criteria 12. There should be evidence that the complete screening programme (test, diagnostic procedures, treatment/intervention) is clinically, socially and ethically acceptable to health professionals and the public.

Genomic testing for specific conditions might meet the criteria. Indeed, non-invasive prenatal screening for Down's syndrome, Patau's syndrome and Edwards' syndrome was approved by the National Screening Committee in 2016. Whether genomic testing for a wide range of gene variants, as a kind of 'fishing exercise', would meet these criteria is unlikely, but in any event would require further research and debate. The consortium of genetic professionals mentioned above state: "We would strongly support the development of well-designed research studies to evaluate the integration of genomic analyses into existing screening programmes, and to develop new programmes to extend the reach of genomic medicine to the prediction of future risk in healthy individuals. The UK is well-placed to deliver such studies and has a strong track record in epidemiology and public health."

Payment by the participant raises a further important issue – that of equity. Offering genomic testing as a paid-for service would mean many people would be excluded from the scheme. This could exacerbate social and health inequalities, with those who are able to afford the test potentially at an advantage over those who cannot afford it.

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