House of Commons

Science and Technology Committee

Direct-to-consumer genomic testing

First Report of Session 2021–22

Report, together with formal minutes relating to the report

Ordered by the House of Commons
to be printed 3 June 2021

HC 94
Published on 22 June 2021
by authority of the House of Commons

Science and Technology Committee

The Science and Technology Committee is appointed by the House of Commons to examine the expenditure, administration and policy of the Government Office for Science and associated public bodies.

Current membership

Greg Clark MP (*Conservative, Tunbridge Wells*) (Chair)

Aaron Bell MP (*Conservative, Newcastle-under-Lyme*)

Dawn Butler MP *(Labour, Brent Central)*

Chris Clarkson MP (*Conservative, Heywood and Middleton)*

Katherine Fletcher MP (*Conservative, South Ribble*)

Andrew Griffith MP (*Conservative, Arundel and South Downs*)

Mark Logan MP (*Conservative, Bolton North East*)

Rebecca Long-Bailey MP *(Labour, Salford and Eccles)*

Carol Monaghan MP (*Scottish National Party, Glasgow North West*)

Graham Stringer MP (*Labour, Blackley and Broughton*)

Zarah Sultana MP (*Labour, Coventry South*)

Powers

The Committee is one of the departmental select committees, the powers of which are set out in House of Commons Standing Orders, principally in SO. No. 152. These are available on the internet via www.parliament.uk.

Publication

Committee reports are published on the Committee's website at www.parliament.uk/science and in print by Order of the House.

Committee staff

The current staff of the Committee are: Masrur Ahmed (Second Clerk), Dr Harry Beeson (Committee Specialist), Dr Christopher Brown (Committee Specialist), Dr James Chandler (Committee Specialist), Emma Dobrzynski (Committee Operations Officer), Sonia Draper (Committee Operations Manager), Danielle Nash (Clerk), and Emily Pritchard (Senior Media and Communications Officer).

Contacts

All correspondence should be addressed to the Clerk of the Science and Technology Committee, House of Commons, London, SW1A 0AA. The telephone number for general inquiries is: 020 7219 2793; the Committee's e-mail address is: scitechcom@parliament.uk.

You can follow the Committee on Twitter using @CommonsSTC.

Contents

Summary

The human genome is the entire sequence of an individual's DNA and is found in almost every cell in the human body. Genomic testing refers to techniques for measuring all or part of this DNA sequence. With the costs of some genomic testing technologies falling significantly in recent years, increasing numbers of genomic testing products have been developed and sold to consumers. These products are typically aimed at providing information about genealogy and ancestry, health, or 'lifestyle' (ranging from dietary advice to earlobe type). One company, 23andMe, had sold over 250,000 genomic testing kits in the UK as of June 2020.

Following a public call for inquiry suggestions, our predecessor Committee launched an inquiry looking at genomic tests sold directly to consumers based on a proposal from the Nuffield Council on Bioethics. We took forward this inquiry. Over the course of our inquiry and our predecessor Committee's inquiry, a range of potential benefits and risks associated with direct-to-consumer genomic testing were raised. These included potential benefits and risks for personal health, opportunities for research and economic growth, considerations of the possible impact on the NHS, and issues related to privacy and consent. Many, but not all, submissions to the inquiries argued that the existing regulations for direct-to-consumer genomic testing should be updated to seize some of these opportunities and address some of the concerns. In its national strategy for genomics, published in September 2020, the Government acknowledged the importance of maintaining public trust in genomics and said that it would "establish a gold standard UK model for how to apply strong and consistent ethical and regulatory standards".

In this Report, we discuss the main opportunities and risks of direct-to-consumer genomic tests in more detail, as well as the available evidence for both. We then present some of the main proposals discussed in the oral and written evidence for seizing opportunities and addressing risks, focusing on testing used for medically related purposes.

Firstly, we recommend that the Government should require direct-to-consumer tests to be subject to greater pre-market assessment by an external body. Currently, most providers are able to self-declare their products' conformity with the existing regulations, which the Medicines and Healthcare products Regulatory Agency suggested restricted its ability to ensure that genomic tests on the UK market provided reliable results. We suggest that any such external assessment should cover the test's clinical performance (the extent to which a test can provide information about diagnosis, treatment, management or prevention of disease that will lead to an improved outcome), as well as its analytical performance (how well a test predicts the presence or absence of a particular gene or genetic change)—which is the focus of requirements on direct-to-consumer genomic tests currently.

Secondly, we propose that the Government should work with Genomics England and the NHS to define clear technical standards for direct-to-consumer genomic testing that, if met, would enable the genomic data generated by the test to be used and trusted by Genomics England and the NHS. The development of such standards, which manufacturers of direct-to-consumer genomic tests could voluntarily meet, could:

reduce the likelihood of false positive or false negative results; facilitate the sharing of data obtained from direct-to-consumer tests, which could potentially support research efforts; and reduce the need for the NHS to re-test individuals following a commercially-obtained test, potentially reducing the burden placed on the NHS by direct-to-consumer tests. They could also provide the means for consumers to discern tests of high quality.

Next, we recommend that the Government should consider the case for amending the current regulation of direct-to-consumer genomic tests to revise the requirements on information and support provided to consumers. This could include requiring companies to inform consumers of the potential consequences of genomic test results for their relatives, or requiring external assessment of the information provided about the tests and results provided, including, for example, studies of consumer understanding. Medical supervision or the provision of genetic counselling could also be required for at least some types of genomic testing offered directly to consumers. The criteria used to determine which tests should require medical supervision could include the severity of the conditions being tested for, as well as the predictive power of the test.

The Government should aim for the data protection framework governing genomic data in the UK to be world-leading. With technologies developing and more consumers using direct-to-consumer genomic tests, existing data safeguards may become less effective and the consequences for privacy more significant. The Government should review the adequacy of the UK's data protection framework for direct-to-consumer genomic testing, including the risks and opportunities presented by technological developments and growing numbers of consumers using direct-to-consumer genomic tests.

We also recommend that the Government should consider if any restrictions should be placed on the types of genomic tests that should be available directly to consumers for use on asymptomatic children or for prenatal testing. For example, the Government may wish to consider banning the provision of genomic tests for use on children that do not meet the criteria of the UK National Screening Committee.

Finally, we recommend that the Government should consider the scope of regulation of direct-to-consumer genomic testing, specifically with respect to companies selling testing products to UK consumers but conducting testing outside of the UK, and companies offering analysis of genomic data obtained by third parties.

1 Introduction

Genomics

The human genome

1. The genome is the entire sequence of an organism's DNA.[1] A human genome contains 3.2 billion 'letters' (building blocks) of DNA and is found in almost every cell in the human body.[2] Around 99.8% of each person's genome is the same as every other person's, but the remaining 0.2% (around 3–4 million letters) is unique to each individual.[3] Genomics England describes the human genome as the "instructions for making and maintaining" a human body.[4] The genome contains the body's genes, short sections of DNA that control the growth and development of the body's cells (the 20,000 genes in the human genome account for around 1% of the total DNA present).[5]

2. Some diseases, such as Huntington's disease and cystic fibrosis, are directly caused by mutations in a single gene.[6] Anyone who carries the relevant gene mutation is at very significant risk of developing the disease (if they have not already).[7] For most common diseases, however, a person's genomic sequence is just one factor that influences the likelihood of developing the condition. Although there are certain genetic factors that can considerably affect the likelihood of developing a disease,[8] the British Society for Genetic Medicine explained that "for most common diseases the genetic contribution is relatively modest and incompletely understood".[9] Other factors such as lifestyle (such as diet or exercise), environmental exposure (such as smoking or exposure to pollutants) and random events (such as the impact of ionising radiation) can have a greater influence over disease susceptibility than genetic factors.[10]

3. Almost all of a person's genome is inherited from their mother and father (some random mutations can occur around the time of conception, causing genetic mutations not inherited from either parent).[11] Some DNA always passes down the maternal line and

1 'The 100,000 Genomes Project', POSTnote 504, Parliamentary Office of Science and Technology, September 2015
2 Department of Health, 'Annual Report of the Chief Medical Officer 2016: Generation Genome' (2017), Chapter 1
3 Department of Health, 'Annual Report of the Chief Medical Officer 2016: Generation Genome' (2017), Chapter 1
4 'What is a genome?', Genomics England, accessed 19 July 2019
5 'Overview: Genetics', NHS, accessed 23 July 2019 and 'The 100,000 Genomes Project', POSTnote 504, Parliamentary Office of Science and Technology, September 2015
6 'Consumer Genetic Testing', POSTnote 407, Parliamentary Office of Science and Technology, March 2012—see also: UK Research and Innovation (CGN0069), paras 2.2–2.3
7 'Monogenic diseases', World Health Organisation, accessed 22 July 2019
8 Dr Ron Zimmern (CGN0020), para 13 and Professor Melinda Mills (CGN0044), para 2.1
9 British Society for Genetic Medicine (CGN0030), section 2
10 See: Micropathology Ltd, University of Warwick (CGN0002); Cancer Genetics Group (CGN0007), para 6; Association of Genetic Nurses and Counsellors (CGN0008); The Royal College of Physicians and the Royal College of Pathologists (CGN0022); PHG Foundation (CGN0023), para 24; British Society for Genetic Medicine (CGN0030), sections 2 and 3; Prenetics International and DNAfit (CGN0035), sections 3 and 7; Clinical Ethics and Law Southampton, University of Southampton (CGN0041), section 1; Professor Melinda Mills (CGN0044), section 2; Dr Pauline McCormack *et al.* (CGN0057), section 5; Dr Elizabeth Ormondroyd (CGN0061), para 1; BioIndustry Association (CGN0068), para 40; UK Research and Innovation (CGN0069), paras 2.2–2.3 and 3.1; 23andMe (COG0002), para 4.5.1
11 NHS, 'Overview: Genetics' and 'Genetic inheritance', both accessed 23 July 2019—mutations can also occur after conception, these would be detectable only in cells derived from the cell in which that original mutation occurred (and may therefore be limited to a particular organ, for example)

some always passes down the paternal line,[12] but the majority of the genome is inherited randomly from each parent's ancestors.[13] With regards to health impacts, some gene sequences do not affect the individual who has them, but may affect their future children.[14] People with such gene sequences are said to be 'carriers' of that genetic condition.

Commercial genomic testing

4. Genomic testing refers to techniques for identifying the content of an individual's DNA, ranging from specific parts of the genome through to whole genome sequencing.[15] Based on the characteristics of the human genome as described in the previous section, genomic tests are typically used for one of three main purposes:

- Genomic tests sold directly to consumers are most commonly used for **genealogical purposes**.[16] Such tests are typically advertised as being able to provide information about genetic ethnicity or to identify existing relatives who have also used the product.[17]

- Genomic testing can be used for a variety of **health-related purposes**. It can be conducted on patients exhibiting signs of disease ("symptomatic" individuals) to provide or confirm a diagnosis, or to guide treatment (if treatment exists).[18] Genomic testing can also be conducted on "asymptomatic" individuals—those not exhibiting any sign of disease. This is usually done to estimate a person's predisposition to developing different conditions in later life. Other medical applications of genomic testing include "carrier testing", which prospective parents can use to assess the risk of passing on an inherited disease, and prenatal testing, which screens fetuses for certain genetic conditions prior to their birth.[19] The NHS has offered a growing variety of genomic tests for many years and plans to continue making increasing use of genomics,[20] but health-related genomic tests are also available for purchase by consumers.[21]

- A third class of genomic tests available to consumers is sometimes known as **'wellness'** tests.[22] These are intended to provide information related to an individual's physical wellbeing or lifestyle. For example, tests are being advertised as providing genetically-tailored diets or exercise plans.[23]

12 Specifically, the Y chromosome is passed down from a father to a son and mitochondrial DNA is always inherited from the mother—International Society of Genetic Genealogy (CGN0010), Annex 1
13 'Principle of independent assortment', Nature Education, accessed 24 July 2019
14 'What can participants find out?', Genomics England, accessed 23 July 2019
15 PHG Foundation (CGN0023), para 10
16 International Society of Genetic Genealogy (CGN0010)
17 For example, see: 'AncestryDNA', Ancestry; 'MyHeritage', MyHeritage; 'Family Finder', Family Tree DNA; 'Full Ancestry Kit', Living DNA—all accessed 12 March 2020
18 National Institute for Health and Care Excellence (CGN0067)
19 PHG Foundation (CGN0023), para 10
20 NHS Health Education England, '70 years of genetics and genomics in healthcare', accessed 16 April 2021 and NHS, 'The NHS Long Term Plan' (2019)
21 For example, see: 23andMe, 'Find out what your DNA says about you and your family'; Dante Labs, 'Whole Genome Sequencing Test: the best lifetime investment you can make for your health and well-being'; Atlas Biomed Group, 'Don't let lockdown get your health down'; Genomics plc, 'Better Healthcare Decisions Powered by Genomics', all accessed 16 April 2021
22 Oral evidence taken on 28 October 2019, HC (2019) 33, Q56
23 For example, see: 'Health Fit', DNAfit; 'What makes you unique?', Orig3n; 'Whole Genome Sequencing Test', Dante Labs; 'Health and Wellbeing Tests', EasyDNA—all accessed 12 March 2020

Regulation

5. A range of different regulations currently apply to genomic tests sold to consumers, including:

- the Consumer Protection Act 1987 and the Consumer Rights Act 2015 (which require products or services sold to consumers to be fit for purpose, as described and meet certain minimum standards, covering aspects such as quality and safety);[24]
- the UK General Data Protection Regulation (which covers the collection, storage and use of data);[25]
- the Human Tissue Act 2004 (which effectively bans DNA analysis without appropriate consent);[26]
- the Advertising Codes (which ban adverts that are misleading, harmful, offensive or irresponsible, and are enforceable under the Consumer Protection from Unfair Trading Regulations 2008 and the Business Protection from Misleading Marketing Regulations 2008);[27] and
- for commercial genomic tests with a medical purpose, the Medical Devices Regulations 2002 (which set out essential requirements for in-vitro diagnostic devices placed on the market, such as requirements on safety for users and for performance to match the manufacturers' claims).[28]

6. Significant changes to the regulation of genomic tests with medical applications were planned by the previous Government. The European Union introduced the new 'in vitro diagnostic medical device' regulation (IVDR) in 2017, which is due to apply fully from May 2022.[29] The previous Parliament passed the Medical Devices (Amendment …) (EU exit) Regulations 2019, which would have caused the UK to adopt essentially the same regulations from May 2022.[30] However, the Government has since introduced the Medical Devices (Amendment …) (EU exit) Regulations 2020 that "revokes the […] IVDR provisions for Great Britain, that would have been implemented" under the previous legislation.[31] As a result of these new regulations, the regulatory framework for commercial

24 Consumer Protection Act 1987 and Consumer Rights Act 2015
25 European Union (Withdrawal) Act 2018, section 3 and Data Protection, Privacy and Electronic Communications (Amendments etc) (EU Exit) Regulations 2019 (SI 2019/419)
26 Human Tissue Act 2004, section 45
27 Committee of Advertising Practice, 'The UK Code of Broadcast Advertising' and 'The UK Code of Non-broadcast Advertising and Direct & Promotional Marketing' (2014)—see also: The Business Protection from Misleading Marketing Regulations 2008 (SI 2008/1276) and The Consumer Protection from Unfair Trading Regulations 2008 (SI 2008/1277)
28 The Medical Devices Regulations 2002 (SI 2002/618) and Directive 98/79/EC of the European Parliament and of the Council
29 Regulation 2017/746 of the European Parliament and of the Council
30 The Medical Devices (Amendment etc.) (EU Exit) Regulations 2019 (SI 2019/791)—the then Government told our predecessor Committee that the UK regulations would "transpose all the key elements contained in the EU IVDR": Department of Health and Social Care and the Department for Business, Enterprise and Industrial Strategy (CGN0053), para 57
31 The Medical Devices (Amendment etc.) (EU Exit) Regulations 2020 (SI 2020/1478), regulation 3 and Schedule 2, para 55; and Explanatory Memorandum to the Medical Devices (Amendment) (EU Exit) Regulations 2020, para 7.19

genomic tests will remain essentially as it is currently, but with the MHRA established as an independent regulatory body outside of the EU framework and provisions made to implement the Northern Ireland Protocol of the Withdrawal Agreement.[32]

Our inquiry

7. In the previous Parliament, our predecessor Committee invited the public to suggest potential inquiries for its future work programme as part of its *My Science Inquiry* work.[33] The Nuffield Council on Bioethics proposed an inquiry into commercial genomics, which was subsequently selected by our predecessor Committee to be taken forward as an inquiry.[34] This inquiry focused on genomic tests sold directly to consumers ('direct-to-consumer genomic tests'), not those used in the healthcare system. This original inquiry ended prematurely as a result of the 2019 General Election, having received over 80 written submissions and after holding two oral evidence sessions.[35]

8. We therefore decided to take forward an inquiry into direct-to-consumer genomic testing, launching a call for additional and updated evidence on 9 April 2020.[36] We published eight pieces of further written evidence and took oral evidence from four witnesses including the Minister for Innovation at the Department of Health and Social Care, Lord Bethell. To assist us in our work, we appointed Professor Frances Flinter, Emeritus Professor of Clinical Genetics at Guy's and St Thomas' NHS Foundation Trust, as a Specialist Adviser for our inquiry.[37] We are grateful to everyone who contributed to our inquiry, as well as to our predecessor Committee's inquiry.

9. In this Report, we review the hopes and concerns surrounding genomic tests sold directly to consumers, as well as the main regulatory changes suggested during the inquiry. Specifically:

- In Chapter 2, we set out the potential benefits and risks of direct-to-consumer genomic tests, and examine the available evidence for either; and
- In Chapter 3, we summarise the proposals made to ourselves and our predecessor Committee for regulatory changes, focusing on medically-related direct-to-consumer tests.

32 Explanatory Memorandum to the Medical Devices (Amendment) (EU Exit) Regulations 2020, paras 2.3–2.7, 6.5–6.6 and 7.9–7.25—see also: HM Government, 'Agreement on the withdrawal of the United Kingdom of Great Britain and Northern Ireland from the European Union and the European Atomic Energy Community' (2019), Protocol on Ireland/Northern Ireland

33 'Committee calls for ideas from the public', House of Commons Science and Technology Committee, accessed 19 July 2019

34 Written evidence submitted to the Science and Technology Committee on 17 December 2018, HC 1716 (MSI0066) and Science and Technology Committee, Sixteenth Report of Session 2017–19, 'My Science Inquiry', HC 1716, para 5

35 House of Commons Science and Technology Committee, 'Commercial genomics inquiry—publications', accessed 12 May 2021

36 House of Commons Science and Technology Committee, 'Science and Technology Committee launches three new inquiries', published 9 April 2020

37 Professor Frances Flinter declared her interests on 24 June 2020: Emeritus Professor of Clinical Genetics, Guy's and St Thomas NHS Foundation Trust; Council Member of the Nuffield Council on Bioethics; Trustee of Alport UK, Progress Educational Trust, Friends of Guy's and St Thomas' Hospitals, Amadeus Chamber Orchestra and European Doctors' Orchestra; occasional work for Guy's and St Thomas' , the Human Fertilisation and Embryology Authority (paid) and scientific journals (unpaid).

2 The benefits and risks of direct-to-consumer genomic testing

10. With the costs of some genomic tests falling significantly in recent years,[38] increasing numbers of genomic testing products have been developed and sold to consumers.[39] For example, one company, 23andMe, had sold over 250,000 genomic testing kits in the UK as of June 2020 (up from 190,000 in April 2019).[40] Over the course of our inquiry and our predecessor Committee's inquiry, a range of potential benefits and risks associated with direct-to-consumer genomic testing were raised. We discuss these in this Chapter.

Potential benefits and risks

Health benefits and risks

11. Many submissions articulated that genomic testing could provide substantial benefits for health, either currently or in the future.[41] A range of health benefits were identified in the evidence, including:

- diagnosing rare genetic conditions that could otherwise take significant time and effort to identify;[42]
- alerting people to their specific health risks, allowing them to potentially mitigate those risks through changed behaviours or personalised screening;[43] and
- indicating the most effective, or least dangerous, treatments for a given patient.[44]

The Human Genetics Unit at the University of Edinburgh—in line with several others—warned, however, that "although knowledge of the health-relevance of genomic information is increasing, it can still only be used for accurate diagnosis and prediction of disease in limited situations" (some of the challenges for its use are discussed in more detail in Chapter 3).[45]

38 Department of Health, 'Annual Report of the Chief Medical Officer 2016: Generation Genome' (2017), Chapter 14

39 'Autosomal Testing Growth', The DNA Geek, accessed 17 February 2020

40 23andMe (CGN0050), para 2.1 and 23andMe (COG0002), para 2.1

41 For example, see: Genomics plc (COG0003); MRC Human Genetics Unit, University of Edinburgh (CGN0006); Cancer Genetics Group (CGN0007), para 1; Academy of Medical Sciences (CGN0021), paras 4–8; PHG Foundation (CGN0023), para 3; British Pharmacological Society (CGN0027), para 5.1; Wellcome Sanger Institute (CGN0039), para 15; Professor Melinda Mills (CGN0044), section 2.2; UK Clinical Genetics Society (CGN0060); Illumina (CGN0063), paras 6.1.1–6.1.6; The Association of the British Pharmaceutical Industry (CGN0066), para 4; National Institute for Health and Care Excellence (CGN0067); UK Research and Innovation (CGN0069), paras 3.2–3.3

42 For example, see: MRC Human Genetics Unit, University of Edinburgh (CGN0006);

43 For example, see: Everything Genetic Ltd (CGN0005); Wellcome Sanger Institute (CGN0039), para 22

44 For example, see: British Society for Genetic Medicine (CGN0030), section 1; Wellcome Sanger Institute (CGN0039), para 22; Congenica Limited (CGN0046), section 4a; Illumina (CGN0063), para 6.1.6; UK Research and Innovation (CGN0069), para 3.2

45 MRC Human Genetics Unit, University of Edinburgh (CGN0006)—see also: PHG Foundation (CGN0023), para 22; Association of Medical Research Charities (CGN0028), para 2; Wellcome Sanger Institute (CGN0039), para 18; Genomics plc (CGN0048);

12. Despite the promise of genomic testing for health benefits, we also heard of potential risks to physical health arising from tests sold directly to consumers. In particular, the possible consequences of consumers receiving inaccurate or misleading results were highlighted:

- consumers overestimating their risk as a result of a direct-to-consumer genomic test could seek unnecessary treatment or avoid particular activities unnecessarily—the British Society for Genetic Medicine gave the example of a woman receiving a false positive result for predisposition to breast cancer, which could lead to her attempting to arrange for an unnecessary mastectomy;[46] and
- consumers underestimating their risk as a result of a direct-to-consumer genomic test could conversely delay seeking medical supervision or persist with risky behaviour due to perceived immunity—the British Pharmacological Society gave the example of an individual deciding to forego a mammogram or a colonoscopy.[47]

The combination of low public understanding, variable test quality and the complexity of correctly interpreting genomic results led many submissions to argue that the chance of results being inaccurate or misinterpreted was high.[48] This is discussed in more detail in Chapter 3.

13. Several submissions also highlighted the potential for direct-to-consumer genomic test results to affect the mental health of consumers.[49] Those taking such tests may, for example, discover that they are predisposed to serious or untreatable disease, or uncover unexpected family relations (such as adoption or misattributed paternity).

Informing parental decisions

14. The PHG Foundation referred to the ability for genomic testing to increase the chance of in-vitro fertilisation leading to a successful pregnancy and/or a baby unaffected by an inherited disease, through screening of eggs, sperm or embryos.[50] Several submissions noted that genomic testing could also help to inform reproductive decisions, for example by:

46 British Society for Genetic Medicine (CGN0030), section 4—see also: PHG Foundation (CGN0023), para 19; British Pharmacological Society (CGN0027), paras 6.1–6.2; Royal College of Physicians of Edinburgh (CGN0036); Macmillan Cancer Support (CGN0051); Dr Pauline McCormack *et al.* (CGN0057), section 3; Dr Elizabeth Ormondroyd (CGN0061), para 4; University of Exeter (CGN0081), para 3.4

47 British Pharmacological Society (CGN0027), para 3.1—see also: Dr Elizabeth Tunbridge (CGN0031), para 3; Congenica Limited (CGN0046), section 4c; Dr Pauline McCormack *et al.* (CGN0057), section 3; University of Exeter (CGN0081), para 3.5

48 For example, see: Cancer Genetics Group (CGN0007), para 4; Shelford Group (CGN0037), para 3; Wellcome Sanger Institute (CGN0039), para 23; Nuffield Council on Bioethics (CGN0049), section 2—see also: Royal College of General Practitioners and British Society for Genetic Medicine, 'Position Statement on Direct to Consumer Genomic Testing' (2019)

49 For example, see: PHG Foundation (CGN0023), para 20; British Society for Histocompatibility and Immunogenetics (CGN0024); Association of Medical Research Charities (CGN0028), paras 21–22; The 'Mind the Risk' consortium (CGN0045), para B(v); Christian Action Research and Education (CGN0054), para 6.9; Genetic Alliance UK (CGN0062), para 7; Mr Darius Meadon (CGN0064); National Institute for Health and Care Excellence (CGN0067); UK Research and Innovation (CGN0069), para 4.3; Biochemical Society (CGN0071), para 1.1; Mrs Debbie Kennett (CGN0073); Antenatal Results and Choices (CGN0075), para 4; Christian Medical Fellowship (CGN0083), section 1

50 PHG Foundation (CGN0023), para 16

- determining if either or both of the parents is a "carrier" for an inherited disease and therefore if a potential child could be at heightened risk of having that disease; or
- determining if a fetus has a genetic condition.[51]

A range of submissions explained that this information could help prospective parents to decide whether or not to try to conceive, to consider the use of pre-implantation genetic diagnosis or prenatal diagnosis,[52] or to continue with a pregnancy and help future parents prepare for a baby with a genetic condition.[53] Indeed, the Clinical Leads of NHS Regional Genetics Services described informing reproductive decisions as one of the main benefits that consumers could derive from genomic testing.[54] Genetic Alliance UK, a charity representing people affected by genetic disease, said that for many individuals with genetic conditions, "reproductive choice" was one of the few "tools available to address the impact of genetic conditions on their lives".[55]

15. Several submissions, however, highlighted ethical issues associated with prenatal genomic testing provided to consumers, in particular its use for informing decisions on terminating pregnancies.[56] Some, such as the Christian Medical Fellowship, argued that it could increase the number of fetuses with genetic conditions that are terminated each year and presented this as an inherently negative outcome.[57] Don't Screen Us Out, a campaign group, told our predecessor Committee that prenatal screening for Down's syndrome had "previously been considered as a way to reduce those being born with Down's syndrome, and can lead to direct conflict with a now well-developed set of human rights and disability rights".[58] Other concerns included that commercial prenatal genomic testing could lead to increased discrimination against, or reduced support for, those suffering from genetic conditions,[59] or facilitate terminations on non-medical grounds, such as gender.[60]

Impact on the NHS

16. Multiple submissions, in particular from companies providing genomic testing to consumers, argued that its use could ease pressure on the NHS, at least in the longer-

51 For example, see: Association of Genetic Nurses and Counsellors (CGN0008); Clinical Leads of NHS Regional Genetics Services (CGN0013); PHG Foundation (CGN0023), paras 10 and 16; British Society for Genetic Medicine (CGN0030), section 1; Wellcome Sanger Institute (CGN0039), para 21; Nuffield Council on Bioethics (CGN0049), section 1; UK Clinical Genetics Society (CGN0060)

52 Pre-implantation genetic diagnosis tests embryos prior to them being implanted into a woman using in-vitro fertilisation, to enable unaffected embryos to be selected for implantation. Prenatal diagnosis can determine if a fetus has a genetic condition or not.

53 See: British Society for Genetic Medicine (CGN0030), section 1; Nuffield Council on Bioethics (CGN0049), section 1; UK Clinical Genetics Society (CGN0060) and Down's Syndrome Association (CGN0085)—see also: Nuffield Council on Bioethics, 'Non-invasive prenatal testing: ethical issues' (2017).

54 Clinical Leads of NHS Regional Genetics Services (CGN0013)—see also: Association of Genetic Nurses and Counsellors (CGN0008)

55 Genetic Alliance UK (CGN0062), para 3

56 For example, see: Mrs Colette Lloyd (CGN0032); Don't Screen Us Out (CGN0034); Royal College of Physicians of Edinburgh (CGN0036); Nuffield Council on Bioethics (CGN0049), section 1; Christian Medical Fellowship (CGN0083), section 5

57 Christian Medical Fellowship (CGN0083), section 5—see also: Don't Screen Us Out (CGN0034), para 7

58 Don't Screen Us Out (CGN0034)

59 For example, see: Mrs Colette Lloyd (CGN0032), section 1(b); Don't Screen Us Out (CGN0034), para 6; Down's Syndrome Association (CGN0085)

60 For example, see: Nuffield Council on Bioethics (CGN0049), section 1; Christian Medical Fellowship (CGN0083), section 5—see also: Nuffield Council on Bioethics, 'Non-invasive prenatal testing: ethical issues' (2017), paras 4.39–4.48

term.[61] For example, Genomics plc told us that consumer testing might ease demand on healthcare systems by facilitating "early intervention and preventive treatment as well as by empowering the individual to take proactive steps to improve their own health and wellbeing".[62] However, a large number of submissions to our inquiry and to our predecessor Committee's inquiry expressed concerns that increased uptake of direct-to-consumer genomic testing could increase pressure on the NHS.[63] The British Society for Genetic Medicine explained:

> Recipients [of results obtained from direct-to-consumer genomic testing] look to the NHS to explore what to do with this information. Additional genetic tests are subsequently performed in NHS laboratories to verify [these] test results […] Surveillance or screening for any risks identified is then often expected from the NHS, yet without appropriately commissioned or NICE recommended pathways to do so.[64]

In addition to the immediate impact of consumers consulting the NHS following a commercial test result, the National Institute for Health and Care Excellence noted that "false negative results could lead to people being diagnosed at a much later stage of disease or living unhealthier lifestyles in the false belief they are at low risk of disease", further adding to pressure on the NHS.[65] The Royal Colleges of Physicians and of Pathologists argued that the current system provided "an uneven playing-field where the commercial providers take profit from offering tests whilst contributing nothing to the NHS providers who are left dealing with unexpected or difficult outcomes".[66]

17. A separate concern was the potential for direct-to-consumer genomic testing to lead to a "two-tier" healthcare system with individuals who could afford such testing obtaining the consequent NHS treatment faster than those who could not afford testing.[67] The British Society of Genetic Medicine explained:

> People who do not qualify for genomic testing under the NHS (because they do not have sufficient risk factors) will, by virtue of their having a [privately-obtained] test result, displace other people who are at higher risk

61 For example, see: 23andMe (COG0002), section 5; Genomics plc (COG0003), paras 9 and 12; Everything Genetic Ltd (CGN0005); Cancer Genetics Group (CGN0007), para 1; Dante Labs SRL (CGN0018); British Society for Histocompatibility and Immunogenetics (CGN0024); Atlas Biomed Group (CGN0029); Prenetics International and DNAfit (CGN0035), section 5; Wellcome Sanger Institute (CGN0039), para 22; National Institute for Health and Care Excellence (CGN0067)

62 Genomics plc (COG0003), para 12

63 For example, see: Roche Products Ltd (COG0004); Micropathology Ltd, University of Warwick (CGN0002); MRC Human Genetics Unit, University of Edinburgh (CGN0006), para 4; Association of Genetic Nurses and Counsellors (CGN0008); Clinical Leads of NHS Regional Genetics Services (CGN0013); The Royal College of Physicians and the Royal College of Pathologists (CGN0022); PHG Foundation (CGN0023), paras 5 and 26–29; British Society for Histocompatibility & Immunogenetics (CGN0024); British Pharmacological Society (CGN0027), paras 2.1 and 8.1; Association of Medical Research Charities (CGN0028), para 14; British Society for Genetic Medicine (CGN0030), section 5; Wellcome Sanger Institute (CGN0039), paras 23 and 31; Wellcome Genome Campus Connecting Science (CGN0040), section (ii); Clinical Ethics and Law Southampton, University of Southampton (CGN0041), sections 2 and 3; Dr Pauline McCormack et al. (CGN0057), section 5; Mr Darius Meadon (CGN0064); Antenatal Results and Choices (CGN0075), para 4; Christian Medical Fellowship (CGN0083), section 5; Dr Susie Cooke (CGN0088)

64 British Society for Genetic Medicine (CGN0030)

65 National Institute for Health and Care Excellence (CGN0067)

66 The Royal College of Physicians and the Royal College of Pathologists (CGN0022)

67 For example, see: The Royal College of Physicians and the Royal College of Pathologists (CGN0022); Clinical Ethics and Law Southampton, University of Southampton (CGN0041), section 3; Nuffield Council on Bioethics (CGN0049), section 1; Dr Elizabeth Ormondroyd (CGN0061), para 4; Genetic Alliance UK (CGN0062), para 18

> of disease. These outcomes could mean that there is inequity of access, with those whose income allows for direct-to-consumer testing being able to 'jump the queue'. Ultimately the provision of commercial genomic testing could lead to greater inequalities in the health system.[68]

Research benefits

18. Several submissions, including from the Academy of Medical Sciences, highlighted the potential for direct-to-consumer genomic testing to contribute to research that could further the fundamental understanding of genomics as well as help to develop new treatments, diagnostics and genomic testing technologies.[69] The European Bioinformatics Institute noted that the larger a genomic dataset, the more useful it becomes for research, as "statistical correlations […] improve".[70] The Wellcome Sanger Institute explained:

> Genomic data holds tremendous value for understanding human biology and disease, but an individual genome only has meaning when compared with many other genomes. The larger the pool of comparison genomes the more accurate the insight. Therefore, the ability for researchers and healthcare professionals to compare large genomic datasets is as important as the ability to generate a dataset. Sharing datasets is crucial for maximising the impact of genomic research, innovation and translation into the NHS.[71]

With many companies offering consumers the option of contributing their genomic data to research projects, direct-to-consumer testing could, submissions to our inquiry argued, potentially provide for larger and more effective databases for research.[72]

Economic opportunity

19. In 2015, a Government-commissioned study estimated that the global genomics market was worth £8 billion—to which the UK industry contributed £800 million—and that it would grow on average 15% per year.[73] In 2017, Arthur D Little, a strategy and innovation consultancy firm, similarly estimated that the global consumer genomic testing market would be $50 billion by 2026.[74] The UK is widely seen as a world-leader in genomic research and application,[75] partly due to strong Government support for the sector, through initiatives such as the 100,000 Genomes Project, the UK Biobank, the

68 British Society for Genetic Medicine (CGN0030), section 5

69 Academy of Medical Sciences (CGN0021), paras 4–5 and 10—see also: 23andMe (COG0002), paras 5.2 and 5.8; Roche Products Ltd (COG0004); P4ML Ltd (CGN0055), section 3; Genetic Alliance UK (CGN0062), para 25; The Association of the British Pharmaceutical Industry (CGN0066), para 2; National Institute for Health and Care Excellence (CGN0067)

70 European Bioinformatics Institute (CGN0038)

71 Wellcome Sanger Institute (CGN0039), para 13

72 See: 23andMe (COG0002), paras 6.4; PHG Foundation (CGN0023), para 17; Prenetics International and DNAfit (CGN0035), section 6; Genetic Alliance UK (CGN0062), para 20; Illumina (CGN0063), paras 3.10

73 Deloitte, 'Genomics in the UK: An Industry Study for the Office of Life Sciences' (2015)

74 Arthur D. Little, 'The advent of consumer owned genetic profiles: Personal genetic services due for explosive growth' (2017)

75 For example, see: MRC Human Genetics Unit, University of Edinburgh (CGN0006); Association of Medical Research Charities (CGN0028), para 5; Wellcome Sanger Institute (CGN0039), para 28; Congenica Limited (CGN0046), section 1; Genomics plc (CGN0048), para 36; 23andMe (CGN0050), para 2.1; Nkaarco Diagnostics Ltd (CGN0058), para 4; Illumina (CGN0063), paras 2.1–2.2; Roche Products Ltd (COG0004)

NHS Genomic Medicine Service and the Life Sciences Sector Deals.[76] Among others, DNAfit, a UK-based direct-to-consumer genomic testing company, highlighted what it saw as a "compelling opportunity for the UK to emerge as a world-class leader in the development and provision of commercial genomic services".[77] Genomics plc, a genomic analysis company spun out from the University of Oxford, argued that "some current and future UK-based small- to medium-sized enterprises could become global players, and in so doing create massive benefits for individuals' health, health care systems, and the economy as a whole".[78]

20. Some expressed concern, however, that the direct-to-consumer genomic testing products and services offered by certain companies provided little genuine value. For example, Professor Timothy Frayling of the University of Exeter argued that "companies selling diet and lifestyle advice tailored to DNA profiles are merely 'sexing up' standard diet and exercise advice to make money from people when there is no evidence of utility of their product".[79]

Privacy and consent

21. In addition to explicit risks and opportunities associated with genomic testing for consumers, submissions also referred to issues related to privacy and consent. Examples included that:

- uses of genomic data may be complicated or unknown at the time the data is collected, with companies asking for permission to use genetic data for broad purposes such as 'research and product development', challenging the principle of informed consent (whereby the person giving consent understands what they are consenting to);[80]
- genomic data may be used for controversial purposes, for example for criminal justice, immigration enforcement, or insurance or job applications;[81]
- genomic samples could be collected and submitted by one individual on behalf of another, without their knowledge or consent;[82] and

76 BioIndustry Association (COG0005), para 6—see also: Genomics England, '100,000 Genomes Project', accessed 11 December 2020; UK Biobank, 'About Us', accessed 11 December 2020; NHS England, 'NHS Genomic Medicine Service', accessed 11 December 2020; HM Government, 'Life Sciences Sector Deal' (2017) and HM Government, 'Life Sciences Sector Deal 2' (2018)

77 Prenetics International and DNAfit (CGN0035), section 2—see also: 23andMe (COG0002), paras 2.1–2.2; Academy of Medical Sciences (CGN0021), paras 12–14; Atlas Biomed Group (CGN0029); Congenica Limited (CGN0046), section 1; Illumina (CGN0063), paras 2.1–2.3; The Association of the British Pharmaceutical Industry (CGN0066), paras 8–9;

78 Genomics plc (COG0003), para 37

79 Professor Timothy Frayling (CGN0080)

80 For example, see: Dr Felicity Boardman (CGN0012), section 3.1; The Royal Society (CGN0019), para 4; British Pharmacological Society (CGN0027), para 7.2; Nuffield Council on Bioethics (CGN0049), section 4; Dr Pauline McCormack *et al*. (CGN0057), section 7b; Mr Darius Meadon (CGN0064)

81 For example, see: Cancer Genetics Group (CGN0007), para 12; Dr Felicity Boardman (CGN0012), section 3.2; Clinical Leads of NHS Regional Genetics Services (CGN0013); Dr Andelka Phillips (CGN0025); Professor Melinda Mills (CGN0044), para 3.3; Congenica Limited (CGN0046), section 4e; Nuffield Council on Bioethics (CGN0049), section 4; Dr Pauline McCormack *et al*. (CGN0057), section 6; UK Clinical Genetics Society (CGN0060)

82 For example, see: Association of Genetic Nurses and Counsellors (CGN0008); British Pharmacological Society (CGN0027), para 7.3; Dr Pauline McCormack *et al*. (CGN0057), section 7a

- commercial collection and storage of data may be vulnerable to security risks, and ownership may change if a company is sold.[83]

22. The Royal Society, however, noted that "many" of these challenges were "not unique, and [were] common across multiple emerging technologies which involve data capture and analysis".[84] The Association of Medical Research Charities noted that one aspect that distinguished direct-to-consumer genomic testing from many other services involving personal data was the potential for test results to have direct relevance to relatives of the person taking the test.[85] The British Society for Genetic Medicine explained that "given sequence data from sufficient people you can predict the sequence data of others":

> For example, if two people in a family provide genomic data, you can deduce that any rare variants present in those two people must also be present in all the relatives who directly connect them.[86]

The ability for distant relatives to be identified from both individuals' genetic data, combined with the growing numbers of people taking genomic tests and making their data publicly available, led some, such as Professor Melinda Mills, Professor of Sociology at the University of Oxford, to highlight the potential for genomic testing to erode privacy:

> Whenever a relative or distant family member makes their data openly available, they unknowingly share data of their kin as well […] Once a genetic database covers around 2% of the population, almost any person could be matched to up to at least a third cousin level (i.e., people who share a great-great-grandparent).[87]

Several submissions argued that this could, for example, lead to criminals, sperm donors or research participants being identified as a result of their distant relatives using a genomic test.[88] Indeed, there has already been one high-profile case of a serial killer in the USA identified using DNA collected from a distant relative for genealogical purposes.[89]

23. The use of direct-to-consumer genomic testing to predict future health risks also raised specific questions in relation to consent and the testing of asymptomatic children.[90] The Nuffield Council on Bioethics explained that "concerns about genetic testing of healthy children often centre on the child's right to an open future and their ability

83 For example, see: University of Oxford (CGN0026), para 3; Nuffield Council on Bioethics (CGN0049), section 4; Mr Darius Meadon (CGN0064); Biochemical Society (CGN0071), para 5.4

84 The Royal Society (CGN0019), para 4

85 Association of Medical Research Charities (CGN0028), para 20—see also: The Royal Society (CGN0019), para 4; British Pharmacological Society (CGN0027), para 7.1; Clinical Ethics and Law Southampton, University of Southampton (CGN0041), section 4; Professor Melinda Mills (CGN0044), para 3.2

86 British Society for Genetic Medicine (CGN0030), section 6

87 Professor Melinda Mills (CGN0044), para 3.3—see also: Curtis Rogers, Partner at GEDmatch.com (CGN0001); The Royal Society (CGN0019), para 4; Shelford Group (CGN0037), para 10; EthicsAndGenetics (CGN0042); Mr Darius Meadon (CGN0064) and Y Erlich *et al.*, 'Identity inference of genomic data using long-range familial searches', Science, vol 362 (2018)

88 For example, see: International Society of Genetic Genealogy (CGN0010), Annex 2; Dr Andelka Phillips (CGN0025); Professor Melinda Mills (CGN0044), para 3.3; Congenica Limited (CGN0046), section 4e; Nuffield Council on Bioethics (CGN0049), section 4; Dr Pauline McCormack *et al.* (CGN0057), section 6; UK Clinical Genetics Society (CGN0060); Mr Darius Meadon (CGN0064); Mrs Debbie Kennett (CGN0073), section 3—see also: and Y Erlich *et al.*, 'Identity inference of genomic data using long-range familial searches', Science, vol 362 (2018)

89 BBC News, 'Golden State Killer suspect traced using genealogy websites', published 27 April 2018

90 For example, see: Shelford Group (CGN0037), para 8; Nuffield Council on Bioethics (CGN0049), section 5; Dr Lucy Frith (CGN0052); Dr Pauline McCormack et al. (CGN0057), section 7b; Ms Kavita Frary (CGN0065), para 3.1

to make their own choices later about accessing their genetic information".[91] Another concern regarding the testing of asymptomatic children, highlighted by researchers at Newcastle University among others, was that tests were being advertised for what some considered to be inappropriate purposes, such as to determine a child's aptitude for sport or arts, with the potential to influence how a child is raised.[92]

Evidence of benefits and risks

24. The previous paragraphs have discussed mostly hypothetical opportunities and risks associated with genomic tests sold directly to consumers. Robust evidence of either, however, appears to be limited. 23andMe, one of the largest direct-to-consumer providers of health-related genomic testing in the world, referred us to the "year-long voluntary post-market surveillance programme" that it had undertaken in 2015 at the request of the Medicines and Healthcare products Regulatory Agency (MHRA):

> 25,360 UK customer results were provided during this period, and 6 reports and meetings were held with the MHRA. During this period, there were no reportable complaints, no reportable incidents and no evidence of a negative impact on the NHS.[93]

However, several submissions described individual occurrences of potential harms arising from genomic testing being offered directly to consumers. Dr Susie Cooke, Head of Medical Genomics at the University of Glasgow, referred to a cancer patient who was considering buying and taking potentially dangerous medication following the results of a commercially obtained test, against the advice of her oncologist.[94] Dr Cooke said that this was an "extreme" but "far from unique" example. The Royal College of Physicians of Edinburgh similarly recounted an example of a family who falsely worried that their child was at risk of heart disease following a genomic test, and therefore thought they should give up competitive sport.[95]

25. Evidence of inaccurate, misleading or poor quality genomic testing being provided to consumers was greater. The British Pharmacological Society cited studies from 2010 and 2013 that found that identical samples submitted to different companies in the USA had led to significantly different results reported back to the consumer.[96] Several submissions highlighted a 2018 study published in *Genetics in Medicine* that found that around 40% of the results reported by some direct-to-consumer testing companies could be incorrect.[97] Researchers at the University of Exeter found that for rare variants (such as those associated with breast and ovarian cancer), more than 80% of positive results reported by some direct-

91 Nuffield Council on Bioethics (CGN0049), section 5

92 Dr Pauline McCormack *et al.* (CGN0057), section 7b—see also: Dr Andelka Phillips (CGN0025); Nuffield Council on Bioethics (CGN0049), section 5; Dr Peter Fotheringham (CGN0082), paras 1–2, 7 and 14–16

93 23andMe (COG0002), para 5.5

94 Dr Susie Cooke (CGN0088)

95 Royal College of Physicians of Edinburgh (CGN0036)

96 British Pharmacological Society (CGN0027), para 4.1—see also: US Government Accountability Office, 'Direct-to-consumer genetic tests: misleading test results are further complicated by deceptive marketing and other questionable practices' (2010) and Kalf *et al.*, 'Variations in predicted risks in personal genome testing for common complex diseases', Genetics in Medicine, vol 16 (2013)

97 For example, see: British Pharmacological Society (CGN0027), para 2.1; Congenica Limited (CGN0046), section 4c; Nuffield Council on Bioethics (CGN0049), section 2; Roche Products Ltd (COG0004)—see also: Tandy-Connor *et al.*, 'False-positive results released by direct-to-consumer genetic tests highlight the importance of clinical confirmation testing for appropriate patient care', Genetics in Medicine vol 20 (2018)

to-consumer tests were false positives.[98] Commenting more generally, DNAfit, a British company providing genomic tests directly to consumers, told our predecessor Committee that the "information and recommendations made by different companies likely differ substantially, even for the same patient", which it described as "probably the biggest issue affecting direct-to-consumer genetic testing today".[99] The Clinical Leads of NHS Regional Genetics Services noted that when the NHS conducted testing on individuals following a commercially-obtained result, the re-testing did not always produce the same results as reported from the direct-to-consumer test.[100] The Association of Genetic Nurses and Counsellors reported the case of a consumer submitting a sample from their dog without this being detected; the company in question did not refute this allegation.[101] Many submissions, including from the British Society for Genetic Medicine, the Academy of Medical Sciences, Atlas Biomed Group and Roche Products, raised concerns over the propensity for health-related genomic tests sold directly to consumers to report false positive, false negative, ambiguous or misleading results.[102] Graeme Tunbridge, Director of Devices at the MHRA, described these broad concerns as "legitimate".[103] Some of the potential reasons for these results, as well as some solutions, are discussed in Chapter 3.

26. Related to the variability of genomic test results from different providers, Dr Susie Cooke, a cancer genomics specialist, also highlighted "inconsistency of what genomic alterations are being tested for between the available tests":

> In a recent survey of eight tests (the majority of which are currently only delivered in the US), each of which reports on several hundred genes in the cancer, only 15% of genes tested were common to all eight assays. This suggests that lots of genes are being tested for which there is no consensus that they are relevant to cancer, opening up a maze of rabbit holes for patients and oncologists to get lost in.[104]

Outside of cancer genomics, Congenica Ltd and the PHG Foundation alleged that one direct-to-consumer tests for Parkinson's disease and susceptibility to breast cancer did not measure all of the genes of relevance to the condition, which the PHG Foundation said had led to the test "fail[ing] to identify nearly 90% of BRCA mutation carriers".[105]

98 University of Exeter (CGN0081), paras 2.1–2.5 and Weedon *et al.*, 'Use of SNP chips to detect rare pathogenic variants: retrospective, population based diagnostic evaluation', British Medical Journal vol 372 (2021)

99 Prenetics International and DNAfit (CGN0035), section 3

100 For example, see: Clinical Leads of NHS Regional Genetics Services (CGN0013); PHG Foundation (CGN0023), para 19; British Society for Genetic Medicine (CGN0030), section 5

101 Association of Genetic Nurses and Counsellors (CGN0008) and Orig3n, Inc. (CGN0077)

102 For example, see: Cancer Genetics Group (CGN0007) para 3; Association of Genetic Nurses and Counsellors (CGN0008); Clinical Leads of NHS Regional Genetics Services (CGN0013); Academy of Medical Sciences (CGN0021), para 15; PHG Foundation (CGN0023), paras 19–20; British Pharmacological Society (CGN0027), para 6.1; Atlas Biomed Group (CGN0029); British Society for Genetic Medicine (CGN0030); Congenica Limited (CGN0046), section 4c; Nuffield Council on Bioethics (CGN0049), section 2; National Institute for Health and Care Excellence (CGN0067); Antenatal Results and Choices (CGN0075), para 4; University of Exeter (CGN0081), paras 2.1–2.5; Roche Products Ltd (COG0004); Letter from Prof Helen Stokes-Lampard to Rt Hon Sir Norman Lamb MP, 7 October 2019

103 Q38

104 Dr Susie Cooke (CGN0088)

105 PHG Foundation (CGN0023), para 19 and Congenica Limited (CGN0046), section 4c—23andMe told us that the information in its Genetic Health Reports to customers informed them that "it is possible to have other genetic risk variants not included in these reports" and that "users could still develop the condition even if they don't have a variant detected": 23andMe (COG0002), para 4.5.1

27. Regarding the potential impact of direct-to-consumer genomic testing on the NHS, 23andMe told us that it had found that under 2% of its UK customers had a specific conversation with their general practitioner about their test result in 2019.[106] In contrast, Dante Labs said that "many [of its customers] took their results to their physician" (although it argued that "none of these people put a burden on their local healthcare systems").[107] Further, multiple organisations representing NHS professionals, such as the British Society for Genetic Medicine, the Association of Genetic Nurses and Counsellors, Clinical Leads of NHS Regional Genetics Services and the Royal Colleges of Physicians and of Pathologists, reported instances of patients seeking support from the NHS after receiving the results of a genomic test procured privately.[108] Antenatal Results and Choices, a charity providing support to families undertaking antenatal screening, said that NHS genetic counselling services were "currently struggling to 'pick up the pieces' when women come to them with unexpected or uncertain results from private [prenatal genomic testing]".[109] The Royal College of General Practitioners told our predecessor Committee that there had been a "lack of academic research on how much private screening results are presented to NHS services and the level of burden this presents", but referenced a 2018 survey of 500 doctors that found that 91% had discussed a private screening result with a patient in an NHS appointment and that just 13% thought that this was a reasonable use of NHS resources (this was not specific to genomic tests).[110]

28. Identifying the lack of robust evidence on the impact of direct-to-consumer genomic testing on the NHS in its inquiry, our predecessor Committee wrote to the then Parliamentary Under-Secretary of State at the Department of Health and Social Care, Baroness Blackwood, recommending that the Government "monitor the impact of direct-to-consumer genomic testing on NHS resources as the use of such tests grows".[111] In response, Baroness Blackwood said that the Government would "work with the NHS and others to explore how to build the evidence base regarding any potential impact and costs to the NHS arising from direct-to-consumer genetic testing".[112] Five months later, the Minister for Innovation at the Department of Health and Social Care, Lord Bethell, told us that:

> Very early exploratory discussion on how such an evidence base could be built have indeed taken place at official level and established that generating robust evidence of impact would require carefully designed survey work to ensure that results were valid.[113]

However, he said that "since those discussions, the Government's priority focus has been to manage the COVID-19 pandemic and therefore this work has so far not been progressed and we have not commissioned or conducted an independent assessment".

106 23andMe (COG0002), para 5.3
107 Dante Labs SRL (CGN0018)
108 See: Association of Genetic Nurses and Counsellors (CGN0008); Clinical Leads of NHS Regional Genetics Services (CGN0013); The Royal College of Physicians and the Royal College of Pathologists (CGN0022); British Society for Genetic Medicine (CGN0030), section 5
109 Antenatal Results and Choices (CGN0075), para 4
110 Letter from Prof Helen Stokes-Lampard to Rt Hon Sir Norman Lamb MP, 7 October 2019
111 Letter from Rt Hon Sir Norman Lamb MP to Baroness Blackwood, 1 November 2019
112 Letter from Baroness Blackwood to the Chair of the Science and Technology Committee, 28 January 2020
113 Lord Bethell, Parliamentary Under Secretary of State for Innovation (COG0009)

The case for updated regulation

29. In light of these concerns, many submissions to our inquiry and our predecessor Committee's inquiry expressed support for updated or strengthened regulation of direct-to-consumer genomic testing, to raise standards, protect consumers and the NHS, and improve consumer trust in the sector.[114] In addition to clinical professional organisations, academics, charities and think tanks, this included companies providing genomic tests for consumers as well as related industry bodies.[115] For example, the Association of the British Pharmaceutical Industry told us that the "development and evolution of a regulatory framework for commercially available tests" was "important, to assure consumers of the validity of the test results".[116]

30. Arguing against updating regulations, Nkaarco Diagnostics, a company providing lifestyle genomic tests directly to consumers, told our predecessor Committee that it was "too late to bring in new regulations to restrict testing".[117] Other companies instead warned against disproportionate regulation.[118] For example, DNAfit said that, while it supported the establishment of a "strict code of conduct to govern the safe applications and interpretation of genomic testing", the Government should "avoid red tape and over-regulation that may limit the ability to foster a sense of discovery and research".[119] Conversely, the Biochemical Society noted that there was an "opportunity for the UK to build a reputation for quality and robustness in genomic testing", and argued that achieving this would "require government regulation to ensure that quality is maintained".[120] The Wellcome Sanger Institute noted that "commercial tests vary greatly in quality and cost and the results are variable among different companies", suggesting that the current lack of regulation or guidance made it difficult for "consumers to distinguish between quality healthcare products and pseudoscientific claims".[121] Dr Matthew Hurles, Head of Human Genetics at the Wellcome Sanger Institute, additionally suggested that "many of the companies would like some regulation" to provide a "bar that they know they need to hit".[122]

31. Several submissions emphasised the different types of genomic tests available, and argued that any regulations should recognise this variety.[123] Conversely, Dr Ron Zimmern,

114 For example, see: The BioIndustry Association (COG0005), para 24; Micropathology Ltd, University of Warwick (CGN0002); Everything Genetic Ltd (CGN0005); Association of Genetic Nurses and Counsellors (CGN0008); Dr Felicity Boardman (CGN0012), section 1.1; The Royal College of Physicians and the Royal College of Pathologists (CGN0022); PHG Foundation (CGN0023), para 7; Dr Andelka Phillips (CGN0025); University of Oxford (CGN0026); Shelford Group (CGN0037), paras 8–9; Congenica Limited (CGN0046), section 1; Genomics plc (CGN0048), para 54; Nuffield Council on Bioethics (CGN0049); Regional Genetics Laboratory (CGN0059), paras 2 and 7; UK Clinical Genetics Society (CGN0060); Dr Elizabeth Ormondroyd (CGN0061), section 5; The Association of the British Pharmaceutical Industry (CGN0066), para 13; Biochemical Society (CGN0071), para 2.3; Down's Syndrome Association (CGN0085) and oral evidence taken on 15 October 2019, HC (2019) 33, Q17

115 For example, see: the BioIndustry Association (COG0005), para 24; Everything Genetic Ltd (CGN0005) and the Association of the British Pharmaceutical Industry (CGN0066), paras 11–14

116 The Association of the British Pharmaceutical Industry (CGN0066), para 13

117 Nkaarco Diagnostics Ltd (CGN0058), para 7

118 For example, see: Prenetics International and DNAfit (CGN0035), section 2 and Genomics plc (CGN0048), paras 53–54

119 Prenetics International and DNAfit (CGN0035), section 2

120 Biochemical Society (CGN0071), para 2.3

121 Wellcome Sanger Institute (CGN0039), para 20—see also: PHG Foundation (CGN0023), para 34

122 Oral evidence taken on 15 October 2019, HC (2019) 33, Q26

123 For example, see: Prenetics International and DNAfit (CGN0035), section 7; Wellcome Sanger Institute (CGN0039), para 14; Genomics plc (CGN0048), paras 53–54; Dr Pauline McCormack et al. (CGN0057), section 7c

Chair of the PHG Foundation, noted that "many […] direct-to-consumer services exist in other forms of medicine" and argued that "genetic services should not be treated any differently":

> Should it be deemed necessary to regulate the direct-to-consumer service sector in health, it should be done across the board. There is no rational reason why genetic services used to predict disease should be regulated in a different way to the use of biochemistry or other modalities of testing.[124]

32. Prior to the Government's decision to revoke aspects of the Medical Devices (Amendment …) (EU exit) Regulations 2019, the UK was due to implement a new regulatory framework for in-vitro diagnostic devices, including genomic tests, from May 2022 (see paragraphs 5–6).[125] The then Government told our predecessor Committee that the new framework that it had planned to introduce would have "drive[n] a much greater scrutiny [of commercial genomic tests] by Notified Bodies".[126] The National Institute for Health and Care Excellence similarly said that these planned regulations "should provide reassurance on a test's reliability and accuracy",[127] while the Nuffield Council on Bioethics told our predecessor Committee that it had "been suggested that [the planned regulations] would be a step towards manufacturers becoming more responsible for the clinical utility of their devices".[128] Graeme Tunbridge, Director of Devices at the Medicines and Healthcare products Regulatory Agency (MHRA), observed that the "amount of work and effort that went into the revision of legislation at EU level pointed to the need to revise what is currently in place".[129] He added that the MHRA had been "heavily involved" in the design of the revised European legislation, and that the "UK was one of the most influential member states when it came to shaping what came out at the end".[130]

33. In its national strategy for genomics, published in September 2020, the Government acknowledged the importance of maintaining public trust in genomics and said that it would "establish a gold standard UK model for how to apply strong and consistent ethical and regulatory standards".[131] However, when it revoked the previously planned strengthening of regulations, the Government committed only to consulting on and publishing future regulations for Great Britain at a "later date".[132]

34. **A range of benefits and concerns have been raised regarding the availability of genomic testing for direct purchase and use by consumers. These apply to all types of genomic tests, but are arguably most acute for tests used for medically-related purposes. Despite concerns around direct-to-consumer tests existing for many years, evidence of harm has mostly not been systematically collected and is limited. As direct-to-consumer genomic testing becomes more widespread and covers a greater variety of conditions, evidence of positive impacts and harms may grow. We have heard calls**

124 Dr Ron Zimmern (CGN0020), para 10
125 See: The Medical Devices (Amendment etc.) (EU Exit) Regulations 2019 (SI 2019/791); The Medical Devices (Amendment etc.) (EU Exit) Regulations 2020 (SI 2020/1478), regulation 3 and Schedule 2, para 55; Explanatory Memorandum to the Medical Devices (Amendment) (EU Exit) Regulations 2020, para 7.19
126 Department of Health and Social Care and the Department for Business, Enterprise and Industrial Strategy (CGN0053), para 54
127 National Institute for Health and Care Excellence (CGN0067)
128 Nuffield Council on Bioethics (CGN0049), section 5
129 Q27
130 Q28
131 HM Government, 'Genome UK: The Future of Healthcare' (2020), p59
132 Explanatory Memorandum to the Medical Devices (Amendment) (EU Exit) Regulations 2020, para 7.16

for updated regulation of direct-to-consumer genomic testing. While the Government has acknowledged the importance of maintaining public trust in genomics and said that it would "establish a gold standard UK model for how to apply strong and consistent ethical and regulatory standards", it has committed only to consulting on and publishing future regulations for Great Britain at a "later date". ***The Government should set out a specific timeframe in which it intends to review the case for introducing new regulations for genomic tests provided directly to consumers ('direct-to-consumer genomic tests').***

3 Supporting a responsible direct-to-consumer genomic testing industry

35. The previous Chapter highlighted the opportunities and risks associated with direct-to-consumer genomic testing, which the Government should seek to seize and mitigate respectively. In this Chapter, we present some of the main proposals discussed in the oral and written evidence for achieving this, focusing on testing used for medically-related purposes.

Continued support for the UK genomics sector

36. The current and previous Governments have implemented a variety of programmes to support genomic testing in the UK.[133] Although these have mostly not focused on genomic tests sold directly to consumers, the BioIndustry Association told our predecessor Committee that the "Government's support instils confidence in the sector and sends strong international signals that the UK continues to be the best location in the world to […] start and grow genomics companies".[134] The latest national strategy for genomics lists some specific measures intended to support the growth of the commercial genomics sector, such as facilitating access to genomic data resources, supporting collaboration and making capital investment available.[135]

37. **Strong support for genomic testing from the current and previous Governments has helped to make the UK a world-leader in the sector. Although this support has focused on testing in the NHS, it has nonetheless supported the direct-to-consumer genomic testing industry. *The Government should continue its support for genomic testing in the UK.***

Ensuring a responsible, trusted genomic testing industry

38. As stated in the previous Chapter, many submissions to our inquiry and to our predecessor Committee's inquiry raised concerns over the risk that some health-related genomic tests sold directly to consumers could report false positive, false negative, ambiguous or misleading results (see paragraph 25).[136] This is despite the current regulation of direct-to-consumer genomic tests under the Medical Devices Regulations 2002, which place obligations on tests including requirements to be "suitable for the purposes […] of providing information concerning a physiological or pathological state, or concerning a congenital abnormality", and to "achieve the performances […] stated

133 For example, see: Genomics England, 'The 100,000 Genomes Project', accessed 4 January 2020; NHS England, 'NHS Genomic Medicine Service', accessed 4 January 2020; HM Government, 'Industrial Strategy: Life Sciences Sector Deal' (2017); HM Government, 'Industrial Strategy: Life Sciences Sector Deal 2' (2018); HM Government, 'Genome UK: The Future of Healthcare' (2020)

134 BioIndustry Association (CGN0068), para 21

135 HM Government, 'Genome UK: The Future of Healthcare' (2020), pp56–58

136 For example, see: Cancer Genetics Group (CGN0007) para 3; Association of Genetic Nurses and Counsellors (CGN0008); Clinical Leads of NHS Regional Genetics Services (CGN0013); Academy of Medical Sciences (CGN0021), para 15; PHG Foundation (CGN0023), paras 19–20; British Pharmacological Society (CGN0027), para 6.1; Atlas Biomed Group (CGN0029); British Society for Genetic Medicine (CGN0030); Congenica Limited (CGN0046), section 4c; Nuffield Council on Bioethics (CGN0049), section 2; National Institute for Health and Care Excellence (CGN0067); University of Exeter (CGN0081), paras 2.1–2.5; Roche Products Ltd (COG0004); Letter from Prof Helen Stokes-Lampard to Rt Hon Sir Norman Lamb MP, 7 October 2019

by the manufacturer".[137] Alleged weaknesses of the current regulations identified during our inquiry and our predecessor Committee's inquiry, as well as some potential remedies, are discussed below.

External validation

39. The Medical Devices Regulations 2002 do not classify genomic tests as a high-risk product (which are subject to more rigorous regulatory approval), allowing most providers to self-declare their products' conformity with the regulations.[138] Graeme Tunbridge, Director of Devices at the Medicines and Healthcare products Regulatory Agency (MHRA), explained that since there was currently "relatively little in the way of pre-market scrutiny" of genomic testing products, it was "incumbent on the company providing it to do what is necessary in making sure that the test is accurate and provides good results in the way they are presented to consumers".[139] He suggested that, whereas currently the MHRA had to "take the company's word" that it was taking appropriate steps to provide products responsibly, regulatory change could require "additional scrutiny by a third party".[140]

40. In line with this, several submissions to our inquiry and our predecessor Committee's inquiry advocated greater scrutiny of companies' products by external organisations.[141] For example, 23andMe, a company that provides medically-related genomic tests directly to consumers, referenced its engagement with the MHRA before and after placing its product on the UK market, and told us that "all new entrants to the market should have discussions about undertaking a similar exercise to ensure that tests are suitable for UK consumers".[142] The Atlas Biomed Group said that by creating an "accessible and respected framework for device and technology registration", the UK could "attract companies, start-ups, investors and scientific talent to biotech hubs around the country".[143]

41. **Most manufacturers of genomic tests sold directly to consumers can self-certify the conformity of their products to performance requirements. The Medicines and Healthcare products Regulatory Agency has suggested that this restricts its ability to ensure that genomic tests on the UK market provide reliable results. *The Government should require manufacturers of direct-to-consumer genomic tests to have the performance of their tests assessed by an external body prior to placing their products on the UK market.***

137 The Medical Devices Regulations 2002 (SI 2002/618), regulation 34—see also: Directive 98/79/EC of the European Parliament and of the Council, Article 1(2)(b) and Annex I (the full text specifies that devices must "achieve the performances, in particular, where appropriate, in terms of analytical sensitivity, diagnostic sensitivity, analytical specificity, diagnostic specificity, accuracy, repeatability, reproducibility, including control of known relevant interference, and limits of detection, stated by the manufacturer")

138 The Medical Devices Regulations 2002 (SI 2002/618), regulations 36 and 40—see also: Directive 98/79/EC of the European Parliament and of the Council, Annexes II, III and IV; British Standards Institute, 'A guide to the In Vitro Diagnostic Directive' (2012); Department of Health and Social Care and the Department for Business, Enterprise and Industrial Strategy (CGN0053), paras 53 and 58–59

139 Q25

140 Q38

141 For example, see: 23andMe (COG0002), para 7.2; Dr Ron Zimmern (CGN0020), para 20; National Institute for Health and Care Excellence (CGN0067)

142 23andMe (COG0002), para 7.2

143 Atlas Biomed Group (CGN0029)

Technical standards

42. Researchers at the University of Exeter suggested that one reason for high rates of false positive results could be the type of testing used by some companies providing genomic testing directly to consumers.[144] The UK Clinical Genetics Society added that it was "not clear whether commercial companies carry out testing to the same high stringent standards that are employed in the NHS, for example to avoid contamination of samples".[145] In line with this, Professor Sir Mark Caulfield, Chief Scientist at Genomics England, told us that whereas the genomic tests performed by Genomics England and provided to the NHS use an "accredited clinical grade pipeline […] some tests done by direct-to-consumer providers meet that standard but some do not".[146] He explained that this meant that Genomics England "could not necessarily take" data obtained from a test provided directly to a consumer and "give it back to the health system as a clinical-grade test":

> That is not to say that the test is invalid, nor that it could not be useful for healthcare. It is simply that, if we are returning direct to patients, we have to have an end-to-end picture of the quality of that data and how it has been gathered.[147]

Professor Caulfield clarified that "for such data to meet the regulatory standards for a clinical grade test the end to end procedure needs to meet ISO accreditation which Genomics England, NHS England and Illumina have attained for the NHS whole genome sequencing programme".[148]

43. Reflecting these concerns, several submissions recommended the development or use of clearer standards for the technical performance of genomic tests, to clarify what was expected and provide assurance that this was being met.[149] Dr Susie Cooke, Head of Medical Genomics at the University of Glasgow, noted that the current framework involved "broad regulations that apply to everything from catheters to MRI scanners", and argued that "what is missing is clear guidance or precedent on how to interpret these regulations to apply them to genomic testing".[150] The PHG Foundation, a non-profit think tank focused on genomics, suggested that "developing some sort of NHS standards to which commercial providers might aspire (similar to encouraging the commercial development of health apps for the NHS Apps library) might allow health professionals

144 University of Exeter (CGN0081), paras 1.1–2.5 and Weedon *et al.*, 'Use of SNP chips to detect rare pathogenic variants: retrospective, population based diagnostic evaluation', British Medical Journal vol 372 (2021)—see also: Cancer Genetics Group (CGN0007), table 1; British Society for Genetic Medicine (CGN0030), sections 4 and 6; Professor Melinda Mills (CGN0044), section 3.2; UK Clinical Genetics Society (CGN0060); NHS Health Education England, 'Consumer genetic testing: expectation and reality', published 15 January 2020

145 UK Clinical Genetics Society (CGN0060)

146 Q68

147 Q68

148 Lord Bethell, Parliamentary Under Secretary of State for Innovation (COG0009), Annex A

149 For example, see: Roche Products Ltd (COG0004); BioIndustry Association (COG0005), paras 24–25; Cancer Genetics Group (CGN0007), table 1; Academy of Medical Sciences (CGN0021), para 26; PHG Foundation (CGN0023), para 38; University of Oxford (CGN0026); Atlas Biomed Group (CGN0029); Shelford Group (CGN0037), para 8; European Bioinformatics Institute (CGN0038) and Dr Susie Cooke (CGN0088) and oral evidence taken on 28 October 2019, HC (2019) 33, Q169

150 Dr Susie Cooke (CGN0088)

to respond to test findings, or even integrate results into NHS patient records".[151] Kathy Hibbs, Chief Legal and Regulatory Officer at 23andMe, added that a "data alignment strategy" could help data-sharing for research purposes.[152]

44. **The results obtained from genomic tests provided directly to consumers cannot be integrated into Genomics England or NHS records since they do not always meet the standards required of the end-to-end testing and data handling process. Enabling tests that are provided directly to consumers to demonstrate that they meet the required standards could help to: reduce the likelihood of false positive or false negative results; facilitate the sharing of data obtained from direct-to-consumer tests, which could potentially support research efforts; and reduce the need for the NHS to re-test individuals following a commercially-obtained test, potentially reducing the burden placed on the NHS by direct-to-consumer tests. *The Government should work with Genomics England and the NHS to define clear technical standards for direct-to-consumer genomic testing that, if met, would enable the genomic data generated by the test to be used and trusted by Genomics England and the NHS. The Government should also establish a mechanism by which providers of direct-to-consumer genomic tests could validate that their tests met these standards.***

Analytic and clinical performance

45. The Medical Devices Regulations 2002 set out the particular attributes of a device that should be used to judge its performance, including its "analytical sensitivity, diagnostic sensitivity, analytical specificity [and] diagnostic specificity".[153] In keeping with this, Dr Ron Zimmern, Chair of the PHG Foundation, argued that the current regulation's "main purpose has been to regulate analytical validity" and that there was "general understanding that it does not, and does not seek to, regulate clinical utility".[154] Illumina, a manufacturer or genomic technologies, explained that:

- analytical validity referred to "how well the test predicts the presence or absence of a particular gene or genetic change"; whereas
- clinical validity referred to "how well the genetic variant being analysed is related to the presence or absence of a phenotype [a physical characteristic, such as a symptom of disease], or risk or predisposition to a specific disease"; and
- clinical utility referred to "whether the test can provide information about diagnosis, treatment, management or prevention of disease that will lead to an improved outcome".[155]

Clinical validity and clinical utility can be considered together as the device's overall 'clinical performance'.

46. Dr Zimmern clarified the importance of both the analytical and clinical properties of a test using an analogy with an X-ray machine, which needs to be safe to use and provide

151 PHG Foundation (CGN0023), para 38—see also: BioIndustry Association (COG0005), para 25; University of Oxford (CGN0026)
152 Oral evidence taken on 28 October 2019, HC (2019) 33, Q107
153 The Medical Devices Regulations 2002 (SI 2002/618), regulation 34—see also: Directive 98/79/EC of the European Parliament and of the Council, Annex I
154 Dr Ron Zimmern (CGN0020), para 30
155 Illumina (CGN0063), para 3.8

the necessary quality of imaging but which also should be used in different ways for different clinical situations and to guide different clinical pathways.[156] Graeme Tunbridge, Director of Devices at the MHRA, suggested that the current requirements were slightly more demanding than those described by Dr Zimmern but nevertheless appeared to agree that they could be strengthened:

> [Under the current regulations,] the provision of clinical evidence alongside the test is relatively limited […] there has to be only a basic analytical correlation between a marker that has been detected and a clinical condition for that to be reported. I think the prevailing view is that you want to see a bit more in the way of clinical validity.[157]

Similarly emphasising the importance of clinical performance, the PHG Foundation said that "harms may arise when a test provider states or implies that a specific test result has certain health implications without robust supporting evidence".[158] The importance of clinical performance, and concerns about the current requirements for clinical performance, were also raised by several others, including the British Society for Genetic Medicine.[159]

47. **The performance requirements on direct-to-consumer genomic tests under the current regulations focus on a genomic test's analytical performance, not its clinical performance. For a medically-relevant test, however, clinical performance is fundamental to how the test will be used by a consumer. *The Government should extend the scope of the performance requirements on direct-to-consumer genomic tests to explicitly cover clinical performance as well as analytical performance.***

48. As indicated by Dr Zimmern's X-ray machine analogy, the clinical performance of a test relies on the interpretation of the test results and what they mean for the individual who was tested. However, multiple submissions emphasised that, even with accurate testing, interpretation of genomic test results to infer information about health was typically complex.[160] Indeed, the British Society for Genetic Medicine stated that its main concern with commercial genomic testing "arises from the juxtaposition of the technical ease and speed of obtaining individual genomic data with the difficulties in interpreting what genomic variation means for an individual and the lack of clear predictions that can be made about future health".[161] The main sources of this complexity outlined in the evidence are discussed below.

156 Dr Ron Zimmern (CGN0020), para 30
157 Q26
158 PHG Foundation (CGN0023), para 18—see also para 35
159 For example, see: British Society for Genetic Medicine (CGN0030), section 7; Professor Melinda Mills (CGN0044), para 3.2; Congenica Limited (CGN0046), sections 1 and 4f; Dr Elizabeth Ormondroyd (CGN0061), para 1
160 For example, see: MRC Human Genetics Unit, University of Edinburgh (CGN0006), para 3; Academy of Medical Sciences (CGN0021), para 15; PHG Foundation (CGN0023), paras 22 and 24; Association of Medical Research Charities (CGN0028), paras 2 and 24; British Society for Genetic Medicine (CGN0030); Shelford Group (CGN0037), paras 3–4; Wellcome Sanger Institute (CGN0039), paras 17–18; Clinical Ethics and Law Southampton, University of Southampton (CGN0041), section 1; Dr Pauline McCormack et al. (CGN0057), section 3; Biochemical Society (CGN0071), paras 3.1–3.2; Christian Medical Fellowship (CGN0083), section 3
161 British Society for Genetic Medicine (CGN0030)

Evidence of clinical performance

49. The first identified source of complexity in assessing the clinical performance of a genomic test was the nature of research used to inform interpretation, which typically looks for variations in the genome that are statistically connected to certain conditions but may not explain the reason for the connection. The National Institute for Health and Care Excellence (NICE) cautioned that there were "thousands of genes in the scientific literature which are associated with disease but that does not mean the presence of the gene has a causal link to the disease or risk of developing disease".[162]

50. Several direct-to-consumer genomic testing companies set out the steps they took to ensure that the results that they returned to their customers were based on reasonable scientific evidence.[163] For example, Kathy Hibbs, Chief Legal and Regulatory Officer at 23andMe, told our predecessor Committee that 23andMe only reported results to consumers that were supported by at least two peer-reviewed studies and, typically, that were included in clinical guidelines.[164] However, some companies also argued that other companies offered tests based on insufficiently robust or transparent evidence.[165] Professor Timothy Frayling of the University of Exeter warned that companies selling tests directly to consumers "clearly have a vested interest in highlighting the handful of research studies that support their claims and ignoring the ones that do not".[166] NICE said that "an independent evidence-based evaluation of the clinical effectiveness of a test could provide reassurance about test accuracy and clinical validity, and ensure that evidence is interpreted appropriately".[167]

51. With regards to the challenge of selecting the appropriate genomic test, Dr Ron Zimmern, Chair of the PHG Foundation, said that it was "important to understand that the individual citizen is not in a position to take a view on the variants which may be clinically beneficial, and that it is professionals who would need to decide on the set of variants (and the genes) that should be analysed".[168] Graeme Tunbridge, Director of Devices at the MHRA, told us that "ensuring that the entire genetic aspect is taken care of" by a test was a "really tough" challenge, noting that "for some diseases there are hundreds or thousands of potential mutations that could have an impact".[169] He suggested that addressing this challenge "almost" required managing tests on a "case-by-case basis".[170]

52. Reflecting the rapidly-developing nature of the scientific understanding underpinning genomic testing, NHS England updates its National Genomic Test Directory annually.[171] Researchers from Newcastle University also told our predecessor Committee that "evidence suggesting relationships between particular genomic markers and diseases or traits has a track record of being modified or even disproven over even short periods of

162 National Institute for Health and Care Excellence (CGN0067)—see also: PHG Foundation (CGN0023), paras 18 and 22
163 For example, see: Prenetics International and DNAfit (CGN0035), sections 3 and 7; 23andMe (COG0002), para 3.7 and Oral evidence taken on 28 October 2019, HC (2019) 33, Qq61–62
164 Oral evidence taken on 28 October 2019, HC (2019) 33, Q61
165 For example, see: Everything Genetic Ltd (CGN0005); Congenica Limited (CGN0046), section 4c
166 Professor Timothy Frayling (CGN0080)
167 National Institute for Health and Care Excellence (CGN0067);
168 Dr Ron Zimmern (CGN0020), para 16
169 Q40
170 Q40
171 NHS England, 'National Genomic Test Directory: Frequently Asked Questions' (2020), p3

time".[172] Graeme Tunbridge told us that a company providing genomic testing "should be constantly updating its clinical evidence" to "ensure it is reflecting the state of the art in medicine".[173]

53. **As the evidence base for genomic testing develops rapidly, some have expressed criticism of the evidence used by some direct-to-consumer genomic testing companies to justify their tests, or warned of the risk of companies 'cherry picking' the most favourable evidence available. Requiring external validation of direct-to-consumer genomic tests, covering clinical as well as analytical performance, could help to address this. *In addition to pre-market validation of direct-to-consumer tests, the Government should consider requiring companies offering such tests to regularly update the evidence submitted to the external validation body, and for that body to review this, for example on an annual basis.***

Medical supervision and genetic counselling

54. The second identified source of complexity in assessing the clinical performance of a genomic test was the variable relevance of the statistical evidence behind a test result for any given individual. Several submissions highlighted that evidence of statistical correlation had often been collected from specific sub-groups of the population (for example those with symptoms of disease or people from specific ethnic backgrounds), which meant that it might not be applicable to the general population or those from other sub-groups, those with or without certain symptoms, or those with certain personal and family medical histories.[174] The British Society for Genetic Medicine emphasised that "interpretation is dependent upon context and clinical evaluation of a patient's symptoms and signs, the family history and the reasons underlying the test request".[175]

55. Related to this challenge of tailoring the interpretation of genomic test results to the individual that used the test, the challenge of selecting the appropriate test to use for a specific individual was also raised. Professor Anneke Lucassen, then Chair of the British Society for Genetic Medicine, explained that "in the health service, if you were looking for a specific genetic disease, you would tend to target the particular gene that you suspected and analyse that in detail".[176] The Cancer Genetics Group warned, however, that if a test

172 Dr Pauline McCormack *et al.* (CGN0057), section 3

173 Q40

174 For example, see: MRC Human Genetics Unit, University of Edinburgh (CGN0006), para 3; Cancer Genetics Group (CGN0007), para 7; PHG Foundation (CGN0023), para 24; British Pharmacological Society (CGN0027), para 5.4; Association of Medical Research Charities (CGN0028), paras 3 and 17; British Society for Genetic Medicine (CGN0030), section 1; Royal College of Physicians of Edinburgh (CGN0036); Wellcome Sanger Institute (CGN0039), para 18; Professor Melinda Mills (CGN0044), para 2.4.1–2.4.2; Genomics plc (CGN0048), para 44; Macmillan Cancer Support (CGN0051); UK Clinical Genetics Society (CGN0060)—As an example of this, Congenica Ltd, a company providing genomic analysis software including to the NHS, pointed to a recent study that found that, whereas a certain gene had previously been associated with a 75% risk of developing diabetes based on its prevalence in families suffering from the disease, data from a broader sample of the population indicated that the risk was more likely under 10%, see: Congenica Limited (CGN0046), section 4c; Wright *et al.*, 'Assessing the Pathogenicity, Penetrance, and Expressivity of Putative Disease-Causing Variants in a Population Setting', The American Journal of Human Genetics, vol 104 (2019)

175 British Society for Genetic Medicine (CGN0030), section 3—see also: The Royal College of Physicians and the Royal College of Pathologists (CGN0022)

176 Oral evidence taken on 15 October 2019, HC (2019) 33, Q6

did not sequence the appropriate genes then a "negative result could be falsely reassuring".[177] Graeme Tunbridge of the MHRA said that ensuring that genomic tests were relevant to the specific individual using them and that they tested all the relevant parts of the genome for a given consumer posed "some very real problems" with "no simple solution".[178]

56. Other challenges raised in the evidence included the relative importance of genetic factors for disease compared to other factors such as lifestyle, environmental exposure and random chance,[179] and the fact that conditions can manifest at different stages in life and with different levels of severity.[180] Many submissions highlighted that these specific challenges were exacerbated by poor public understanding of genomic testing.[181] A research group at the University of Southampton, for example, highlighted that the reality of complex and often uncertain interpretation "strongly contrasts with the public image that exists around genetics, which is [that it is] overwhelmingly clear-cut and unambiguous in terms of the ability to predict future health risks".[182]

57. Many submissions highlighted how the challenges discussed in paragraphs 54 to 56 were managed in the NHS, and contrasted this with the practices of direct-to-consumer genomic testing. The British Society for Genetic Medicine, for example, stated that NHS genetic services "utilise family history and clinical information to interpret results", whereas "commercial testing often does not take any contextual factors into account, meaning that the chance of false positive and false negatives is much higher".[183] The Association of Genetic Nurses and Counsellors warned that "commercially available genetic tests do not tailor-make the testing to a specific family history of disease, nor [the patient's clinical symptoms]", meaning that the "appropriate test may not be done".[184] The Shelford Group of research-intensive NHS trusts similarly highlighted the "huge difference between testing a symptomatic person for clinical reasons to find a diagnosis and formulate a management plan for that person and relevant relatives within the NHS versus doing a massive test for non-specific reasons".[185] Finally, the Cancer Genetics Group told our predecessor Committee that "without the interpretative support of a genetic counsellor [as

177 Cancer Genetics Group (CGN0007), para 3—see also: Association of Genetic Nurses and Counsellors (CGN0008); PHG Foundation (CGN0023), para 19; British Pharmacological Society (CGN0027), para 6.1; Congenica Limited (CGN0046), section 4c; Nuffield Council on Bioethics (CGN0049), section 2; Royal College of General Practitioners and British Society for Genetic Medicine, 'Position Statement on Direct to Consumer Genomic Testing' (2019)

178 Q40

179 British Society for Genetic Medicine (CGN0030), section 2—see also, for example: The Royal College of Physicians and the Royal College of Pathologists (CGN0022); PHG Foundation (CGN0023), para 24; Association of Medical Research Charities (CGN0028), para 2; Clinical Ethics and Law Southampton, University of Southampton (CGN0041), section 1; Professor Melinda Mills (CGN0044), para 2.1; Dr Elizabeth Ormondroyd (CGN0061), para 1; UK Research and Innovation (CGN0069), para 4.2.1; Biochemical Society (CGN0071), para 3.1

180 Dr Felicity Boardman (CGN0012), section 3.3—see also: Cancer Genetics Group (CGN0007), para 6; Association of Medical Research Charities (CGN0028), para 4

181 For example, see: British Society for Histocompatibility and Immunogenetics (CGN0024); British Pharmacological Society (CGN0027), para 2.1; Association of Medical Research Charities (CGN0028), para 24; British Society for Genetic Medicine (CGN0030); Shelford Group (CGN0037), paras 3–4; Wellcome Sanger Institute (CGN0039), para 17; Wellcome Genome Campus Connecting Science (CGN0040), section (ii); Clinical Ethics and Law Southampton, University of Southampton (CGN0041); Professor Melinda Mills (CGN0044), para 4.1; The 'Mind the Risk' consortium (CGN0045), para B(iii); Dr Pauline McCormack *et al.* (CGN0057), sections 1, 3 and 5; Dr Elizabeth Ormondroyd (CGN0061), para 1; Christian Medical Fellowship (CGN0083), section 3; Down's Syndrome Association (CGN0085)

182 Clinical Ethics and Law Southampton, University of Southampton (CGN0041)

183 British Society for Genetic Medicine (CGN0030)

184 Association of Genetic Nurses and Counsellors (CGN0008)—see also: Cancer Genetics Group (CGN0007), para 3; British Society for Genetic Medicine (CGN0030), section 1

185 Shelford Group (CGN0037)—see also: Dr Ron Zimmern (CGN0020), para 11; Association of Medical Research Charities (CGN0028)

would be provided in the NHS], a significant proportion of the population may not fully understand percentages or relative risks compared to the general population and therefore misinterpret their results".[186] The Association of Genetic Nurses and Counsellors said that the counselling and support services offered by most direct-to-consumer genomic testing companies was "severely lacking".[187]

58. In line with the personalised testing and interpretation provided by the NHS to ensure appropriate testing and interpretation, some submissions suggested that at least some genomic tests should only be available to consumers through a medical intermediary.[188] The Royal Colleges of Physicians and of Pathologists drew comparison with pharmaceuticals that are approved for 'over the counter' sale or on prescription only, and advised that "genomic tests could be approved for 'access via a registered health professional' or 'direct to consumer' use".[189] The Wellcome Sanger Institute made a similar recommendation.[190] A strict form of this approach has been adopted by a variety of European countries, including France, Germany and Spain, which require medical supervision for all health-related genomic tests.[191] The Royal College of General Practitioners made a related suggestion that regulations should require companies providing genomic testing directly to consumers to "provide clinical support" to their customers.[192]

59. Closely related to calls for medical supervision, over 30 submissions to our inquiry and our predecessor Committee's inquiry stated the importance of adequate genetic counselling alongside testing,[193] and at least fourteen explicitly recommended that some

186 Cancer Genetics Group (CGN0007), para 3—see also: British Society for Histocompatibility and Immunogenetics (CGN0024); Atlas Biomed Group (CGN0029); Regional Genetics Laboratory (CGN0059), para 4

187 Association of Genetic Nurses and Counsellors (CGN0008)—see also: Clinical Leads of NHS Regional Genetics Services (CGN0013); Mrs Colette Lloyd (CGN0032), section 1(a); Wellcome Genome Campus Connecting Science (CGN0040), section (i); Clinical Ethics and Law Southampton, University of Southampton (CGN0041), section 2; Nuffield Council on Bioethics (CGN0049), section 3

188 For example, see: The Royal College of Physicians and the Royal College of Pathologists (CGN0022); Wellcome Sanger Institute (CGN0039), para 12; Dr Pauline McCormack *et al.* (CGN0057), section 4; Regional Genetics Laboratory (CGN0059), para 4; Genetic Alliance UK (CGN0062), para 15

189 The Royal College of Physicians and the Royal College of Pathologists (CGN0022)

190 Wellcome Sanger Institute (CGN0039), para 12

191 Kalokairinou *et al.*, 'Legislation of direct-to-consumer genetic testing in Europe: a fragmented regulatory landscape', Journal of Community Genetics, vol 9 (2018)

192 Letter from Prof Helen Stokes-Lampard to Rt Hon Sir Norman Lamb MP, 7 October 2019

193 For example, see: Micropathology Ltd, University of Warwick (CGN0002); Simon Davies (CGN0003); Everything Genetic Ltd (CGN0005); Cancer Genetics Group (CGN0007), para 4; Association of Genetic Nurses and Counsellors (CGN0008); Dr Felicity Boardman (CGN0012), section 1.1; Clinical Leads of NHS Regional Genetics Services (CGN0013); Dr Ron Zimmern (CGN0020), para 10; PHG Foundation (CGN0023), paras 7, 23 and 35; British Society for Histocompatibility and Immunogenetics (CGN0024); British Pharmacological Society (CGN0027), paras 7.2 and 7.5; Atlas Biomed Group (CGN0029); British Society for Genetic Medicine (CGN0030), section 7; Mrs Colette Lloyd (CGN0032), section 1(a); Prenetics International and DNAfit (CGN0035), section 4; Royal College of Physicians of Edinburgh (CGN0036); Shelford Group (CGN0037), para 8; Wellcome Genome Campus Connecting Science (CGN0040), section (i); Clinical Ethics and Law Southampton, University of Southampton (CGN0041), section 2; The 'Mind the Risk' consortium (CGN0045), para C(i); Congenica Limited (CGN0046), section 1; Genomics plc (CGN0048), para 53; 23andMe (CGN0050), paras 4.4 and 4.6.1; Christian Action Research and Education (CGN0054), para 6.4; Dr Pauline McCormack et al. (CGN0057), section 4; Regional Genetics Laboratory (CGN0059), para 4; UK Clinical Genetics Society (CGN0060); Genetic Alliance UK (CGN0062), paras 15–16; Ms Kavita Frary (CGN0065), para 4.3; National Institute for Health and Care Excellence (CGN0067); Biochemical Society (CGN0071), paras 4.1–4.4; Down's Syndrome Association (CGN0085)

form of genetic counselling be required for at least some direct-to-consumer genomic tests.[194] The Association of Genetic Nurses and Counsellors explained that genetic counselling involved supporting individuals through the genomic testing process, including:

- explaining the potential outcomes of tests to individuals before they take the test, and helping to order the appropriate test;
- supporting interpretation of test results and explaining the significance and potential options for individuals after a test;
- referring the individual for appropriate treatment or screening following a test; and
- managing the psychological and ethical aspects of returning test results, including mediating consequent testing of family members.[195]

Professor Anna Middleton, Chair of the Association of Genetic Nurses and Counsellors, emphasised that genetic counselling was different from more general therapeutic counselling.[196]

60. Arguing against a requirement for genetic counselling with direct-to-consumer genomic tests, Kathy Hibbs, Chief Legal and Regulatory Officer for 23andMe, said that 23andMe's product had "been designed to be sold over the counter without provision" of counselling, and that "numerous user comprehension studies" had "demonstrated that it is safe and effective without the counselling".[197] However, despite its confidence in the information it provides to its customers, 23andMe recommends that "users speak with a genetic counsellor before testing, and also after testing, to help them understand their results and what actions they should take".[198] Ms Hibbs explained that there "were times when [23andMe] offered genetic counselling", but "only a tiny percentage of people ever utilised it", and that it "would probably be inappropriate" for 23andMe to provide counselling itself.[199] She added that US studies had found that "as many as 40% of women prescribed a BRCA test by their physician […] never actually utilised the test if genetic counselling was required pre-test", which she argued meant that there were "access issues that could be driven by imposing a counselling requirement as well" (although she noted that this issue "could be different in the UK").[200]

194 For example, see: Everything Genetic Ltd (CGN0005); Cancer Genetics Group (CGN0007), table 1; Association of Genetic Nurses and Counsellors (CGN0008); Dr Ron Zimmern (CGN0020), para 10; PHG Foundation (CGN0023), paras 23 and 35; British Society for Genetic Medicine (CGN0030), section 7; Royal College of Physicians of Edinburgh (CGN0036); Shelford Group (CGN0037), para 8; Wellcome Genome Campus Connecting Science (CGN0040), section (i); Genomics plc (CGN0048), para 53; Dr Pauline McCormack et al. (CGN0057), section 4; Regional Genetics Laboratory (CGN0059), para 4; Biochemical Society (CGN0071), para 6.1; oral evidence taken on 15 October 2019, HC (2019) 33, Q15

195 Association of Genetic Nurses and Counsellors (CGN0008)

196 Q27

197 Oral evidence taken on 28 October 2019, HC (2019) 33, Qq84, 120 and 127—see also: letter from Kathy Hibbs to the Science and Technology Committee, 14 January 2020

198 23andMe (COG0002), para 4.6.1

199 Oral evidence taken on 28 October 2019, HC (2019) 33, Qq119–129

200 Oral evidence taken on 28 October 2019, HC (2019) 33, Q129

61. Some submissions suggested that genetic counselling should be offered with any genomic test sold directly to consumers.[201] The Human Genetics Commission, a former advisory body to the UK Government, also recommended in 2010 that pre- and post-test counselling be offered with any "genetic test in the context of inherited or heritable disorders".[202] The European Society of Human Genetics and the American College of Medical Genetics and Genomics have each recommended that expert advice, tailored to the specific person using the test, be provided to consumers before and after using health-related genomic tests.[203] However, Professor Anna Middleton, Chair of the Association of Genetic Nurses and Counsellors, told our predecessor Committee that she did not think that "genetic counselling *per se* should be the gatekeeper for the ability to access a genetic test", because the need for it was "context specific".[204] She recommended instead that genetic counselling should be "offered pre- and post-test for serious, life-threatening, inherited conditions".[205] Professor Anneke Lucassen, then Chair of the British Society for Genetic Medicine, similarly stated that the Society was not "calling for counselling for all genetic tests", but thought that it should be provided with tests for all "very high-risk conditions" or for "highly predictive test[s]" with "big implications for either no treatment or drastic treatment".[206] Several other submissions, such as from the PHG Foundation and the Royal College of Physicians of Edinburgh, made similar recommendations.[207] The Nuffield Council on Bioethics recommended that counselling be required prior to and after non-invasive prenatal testing, a form of genomic testing for fetuses during pregnancy.[208] Genomics plc, a company offering genomic tests to consumers through healthcare providers, argued against requirements for genomic counselling where tests provided information related to common diseases by detecting large numbers of genetic factors each with weak influence on the overall risk, saying that the "information communicated in those cases would refer to individually small effects on risk for individuals and their families" and should be treated like predictive tests for other risk factors such as blood pressure or cholesterol.[209]

62. The changes to regulation planned by the previous Government would have introduced a requirement for "appropriate access to counselling" to be provided if the genomic test pertained to "genetic predisposition for medical conditions or diseases which are generally considered to be untreatable".[210] Professor Middleton described this focus on untreatable conditions as an "unhelpful distinction", noting that treatable conditions could still require management of appropriate screening measures and interaction with relatives, entailing an "awful lot of thought and consideration of the family dynamics and

201 For example, see: Wellcome Genome Campus Connecting Science (CGN0040), section (i); Dr Pauline McCormack et al. (CGN0057), section 4; Nkaarco Diagnostics Ltd (CGN0058), para 6; Genetic Alliance UK (CGN0062), para 15; Biochemical Society (CGN0071), paras 3.2 and 4.1–4.4

202 Human Genetics Commission, 'A Common Framework of Principles for direct-to-consumer genetic testing services' (2010), p9

203 Q15—see also: European Society of Human Genetics, 'Statement of the ESHG on direct-to-consumer genetic testing for health-related purposes' (2010) and American College of Medical Genetics and Genomics, 'Direct-to-consumer genetic testing: a revised position statement of the American College of Medical Genetics and Genomics' (2016)

204 Q28

205 Oral evidence taken on 15 October 2019, HC (2019) 33, Q49

206 Oral evidence taken on 15 October 2019, HC (2019) 33, Qq29 and 35

207 For example, see: Q46; Dr Ron Zimmern (CGN0020), paras 10 and 37; PHG Foundation (CGN0023), para 23; Royal College of Physicians of Edinburgh (CGN0036); Genomics plc (CGN0048), para 53

208 Nuffield Council on Bioethics, 'Non-invasive prenatal testing: ethical issues' (2017), para 6.40

209 Genomics plc (CGN0048), para 53—'monogenic' diseases

210 The Medical Devices (Amendment etc.) (EU Exit) Regulations 2019 (SI 2019/791), regulation 11

the moment when you need to test, and loads of counselling can go with that".[211] Graeme Tunbridge of the MHRA similarly told us that the previously planned regulatory changes for counselling were a "fudge".[212]

63. Professor Middleton also noted that the regulation was ambiguous on what would constitute appropriate counselling.[213] She argued that "counselling has to be a two-way process; it is a dialogue and it is patient-centred, because it is focused on what the patient needs", and clarified that support for consumers delivered through written information or using automated systems "would not be counselling".[214] The UK Clinical Genetics Society, a professional body, similarly noted that "patients accessing genetic testing through the NHS are usually offered a face to face appointment", which it said "may be an essential component of their evaluation as it may enable a clinical diagnosis to be made".[215] In its written evidence, the Association of Genetic Nurses and Counsellors also argued that counselling should be delivered by "trained and regulated genetic counsellors".[216] Several other submissions agreed on the importance of professionally qualified counsellors.[217]

64. **Results obtained from genomic testing must typically be considered in the context of an individual's specific circumstance—including their symptoms, personal and family medical history and ethnicity—in order for the clinical significance of those results to be interpreted correctly. These personal details are also often pertinent to the selection of the most appropriate genomic test for an individual. The NHS takes account of these contextual factors through the expert supervision and counselling provided throughout the testing process. In contrast, companies providing genomic testing directly to consumers appear to provide generic tests and depend upon consumers understanding written information provided alongside the test that has not been tailored to their personal situation.** ***The Government should consider the case for amending the regulation of genomic tests provided directly to consumers, to require medical supervision or the provision of genetic counselling for at least some types of genomic testing offered directly to consumers. Criteria used to determine which tests should require medical supervision could include the severity of the conditions being tested for, as well as the predictive power of the test. Requirements for supervision and genetic counselling should cover the qualifications of the medical intermediary required and minimum requirements on the content and format of the support or oversight provided.***

Information for consumers

65. As described in paragraph 60, 23andMe argued—despite recommending that its customers consult a genetic counsellor—that the information provided with its test results made them safe without counselling.[218] The updated regulations planned by the previous

211 Oral evidence taken on 15 October 2019, HC (2019) 33, Q49
212 Q33
213 Oral evidence taken on 15 October 2019, HC (2019) 33, Q45
214 Oral evidence taken on 15 October 2019, HC (2019) 33, Q29
215 UK Clinical Genetics Society (CGN0060)—see also: oral evidence taken on 15 October 2019, HC (2019) 33, Q29
216 Association of Genetic Nurses and Counsellors (CGN0008)
217 For example, see: Prenetics International and DNAfit (CGN0035), section 4; Royal College of Physicians of Edinburgh (CGN0036); Wellcome Genome Campus Connecting Science (CGN0040), section (i); Dr Pauline McCormack et al. (CGN0057), section 4; Regional Genetics Laboratory (CGN0059)
218 Oral evidence taken on 28 October 2019, HC (2019) 33, Qq84, 120 and 127—see also: letter from Kathy Hibbs to the Science and Technology Committee, 14 January 2020; 23andMe (COG0002), paras 3.8–3.9 and 4.6–4.12

Government would have introduced a requirement for users of medically-relevant genomic tests to be "provided with relevant information on the nature, the significance and the implications of the genetic test".[219] Graeme Tunbridge of the MHRA suggested that updated regulation could provide for additional external assessment of the information provided to consumers along with their test results:

> [External scrutiny by a third party could cover] much more in the way of data to make sure [it was] happy that when [a provider is] reporting something it is reported properly, that the risk of failure is properly reported as well and that the consumer can properly understand the result based on all those factors.[220]

66. Professor Anneke Lucassen, then Chair of the British Society for Genetic Medicine, acknowledged that "certain companies have excellent websites" and said that the "problem lies more in the expectations of the people using the test".[221] She suggested that, for this reason, she "might say that it is not so much about regulating the companies as about improving the information around what a genetic test might and might not tell you".

67. Related to this point, several submissions expressed concern regarding the advertising of direct-to-consumer genomic tests.[222] Dr Felicity Boardman of Warwick Medical School told our predecessor Committee that research had "consistently found that advertising of tests [offered directly to consumers] typically falls short of the information standard that would be expected of testing provided within state funded healthcare settings".[223] She said that "overstatement of the effectiveness of the tests, both in terms of their analytic and clinical utility" was "not uncommon", nor was a "minimisation of the risks associated with uncovering this knowledge, in terms of impacts for biological relatives as well as mental and physical health".[224] The Royal College of General Practitioners warned that misleading advertising of direct-to-consumer genomic tests "feeds unrealistic patient expectations of healthcare", with researchers at Newcastle University noting that "consumers are likely to assume that genetic testing […] is always highly predictive and determinative".[225]

68. Professor Anna Middleton, Chair of the Association of Genetic Nurses and Counsellors, explained that one problem was the contrast between the marketing for tests that "implies that you can use them clinically" and the "small print [where] it is very clear that they are not to be used clinically".[226] Another problem identified by several submissions concerned the statistics used in advertisements for genomic tests. For example, the Nuffield Council on Bioethics warned that some commercial providers were engaging in "misleading use of statistics and language about the accuracy of non-

219 The Medical Devices (Amendment etc.) (EU Exit) Regulations 2019 (SI 2019/791)—see also: Regulation 2017/746 of the European Parliament and of the Council, Article 4

220 Q38—see also: Q40

221 Oral evidence taken on 15 October 2019, HC (2019) 33, Q13

222 For example, see: Dr Felicity Boardman (CGN0012), section 3.3; British Society for Genetic Medicine (CGN0030), section 2; Mrs Colette Lloyd (CGN0032); Professor Melinda Mills (CGN0044), section 3.2; Nuffield Council on Bioethics (CGN0049), section 5; Dr Pauline McCormack *et al.* (CGN0057), section 3; Down's Syndrome Association (CGN0085); letter from Prof Helen Stokes-Lampard to Rt Hon Sir Norman Lamb MP, 7 October 2019

223 Dr Felicity Boardman (CGN0012), section 1.1

224 Dr Felicity Boardman (CGN0012), section 3.3

225 Letter from Prof Helen Stokes-Lampard to Rt Hon Sir Norman Lamb MP, 7 October 2019 and Dr Pauline McCormack *et al.* (CGN0057), section 3

226 Oral evidence taken on 15 October 2019, HC (2019) 33, Q15

invasive prenatal testing".[227] It explained that companies were often highlighting high negative predictive values (the proportion of the fetuses that test negative who are actually unaffected) without making clear that the positive predictive values (the proportion of the fetuses that test positive who are actually affected) could be significantly lower.[228] Dr Rachel Horton of the University of Southampton has noted that low false negative rates can also be misleading:

> Generally, direct-to-consumer genetic testing companies' reports specify if they have only taken a very limited look at particular genes, and some have really impressive and detailed information about this on their websites. However, the stats they give about false negatives understandably relate to the test they're actually doing: so a reported false negative rate below 1% means that, for the particular variations within a gene that a test looks at, the test would miss less than 1% of people who have one of these exact variations. However, a BRCA test only looking at three common variants would miss around 80% of people with a BRCA variant that increased their chance of cancer.[229]

69. Amidst these concerns, the Royal College of Physicians of Edinburgh said that it was "vital that advertising of direct to consumer genetic tests is truthful and describes the offer in intelligible language, with easily understandable information about the limitations of the tests on offer as well as the benefits".[230] The British Pharmacological Society added that companies should "provide a clear picture of how comprehensive their test or interpretation is", in particular by specifying which variants were and were not tested for.[231] The Royal Colleges of Physicians and Pathologists gave examples of the information that they thought companies should provide with their tests, including:

- the scope of the test;
- who the test is intended for;
- the potential benefits and harms of the test;
- the sensitivity and specificity of the test;
- the likely range of results from the test; and
- the recommended strategies for managing the results.[232]

70. In 2020, the Advertising Standards Authority issued three rulings against commercial providers of non-invasive prenatal testing, determining that their advertisements were misleading (although the statistics they had quoted were accurate) because consumers

227 Nuffield Council on Bioethics (CGN0049), section 1—see also: Mrs Colette Lloyd (CGN0032), section 1a; Don't Screen Us Out (CGN0034), para 6; Dr Pauline McCormack et al. (CGN0057), section 7b; Antenatal Results and Choices (CGN0075), para 3

228 Nuffield Council on Bioethics, 'Non-invasive prenatal testing: ethical issues' (2017), paras 1.19, 2.23 and 4.22

229 NHS Health Education England, 'Direct-to-consumer testing: a clinician's guide', accessed 11 May 2021—BRCA refers to certain genes, mutations in which have been associated with an increased risk of developing breast cancer

230 Royal College of Physicians of Edinburgh (CGN0036)

231 British Pharmacological Society (CGN0027), para 4.2

232 The Royal College of Physicians and the Royal College of Pathologists (CGN0022)

were likely to misunderstand what the statistics meant.[233] It advised that, going forwards, "providers are best advised to stay away from using 'detection rates' in their ads as they can confuse and mislead consumers", and that "if providers do want to use them, they should also provide the 'positive predictive value' percentage alongside a clear explanation of what both figures actually mean to avoid misleading consumers". The Committee of Advertising Practice issued an enforcement notice setting out these guidelines, contacted providers to ensure that they were complying with the new guidelines and stated that it would refer any companies that refused to comply to Trading Standards or relevant professional regulatory bodies.[234] Looking beyond non-invasive prenatal testing alone, the Nuffield Council on Bioethics has recommended that the "responsible authorities pay more attention to whether genetic test providers are making clinical claims for their products, even if implied rather than explicit".[235]

71. **Several contributors to this inquiry expressed concern that the information provided to consumers before and after using a direct-to-consumer genomic test, as well as the advertising used to market direct-to-consumer genomic tests, did not do enough to address public misconceptions of the capability of these tests and clarify the clinical utility of the results generated. Even where advertising material has used statistics accurately, the Advertising Standards Authority has ruled that there is still scope for them to provide a misleading impression to consumers. *The Government should consider the case for including reviews of the information provided to consumers prior to and after taking a direct-to-consumer test within any external validation required to place such tests on the market. This could, for example, include assessment of studies of consumer understanding of the information provided.***

72. ***Building on its review of advertising for non-invasive prenatal testing, the Advertising Standards Authority should review, within the next year, the marketing materials used by companies offering other genomic tests directly to consumers, focusing in particular on the clinical performance implied by the tests compared with their actual performance.***

73. Several submissions criticised the level of support offered to consumers before and following a non-invasive prenatal test in particular.[236] Antenatal Results and Choices, a charity providing information and support to those undertaking antenatal screening, highlighted that "results from non-invasive prenatal testing can lead to profound decisions for expectant parents", with Don't Screen Us Out, a campaign group, noting that "90% of pregnant women who screen for Down's syndrome go on to terminate the pregnancy".[237] Agreeing that there was "frequently a presumption that termination will be the next choice that [prospective parents] will make" following a positive prenatal test, the Down's Syndrome Association therefore highlighted the importance of "up to date, balanced and accurate information about the condition being screened for and counselling to

233 Advertising Standards Authority, 'Non-Invasive Prenatal Testing (NIPT)—A look at the ASA's rulings', published 16 January 2020

234 Committee of Advertising Practice, 'Enforcing the rules for ads on prenatal testing', published 16 January 2020 and Advertising Standards Authority, 'Non-Invasive Prenatal Testing (NIPT)—A look at the ASA's rulings', published 16 January 2020

235 Nuffield Council on Bioethics (CGN0049), section 5

236 For example, see: Mrs Colette Lloyd (CGN0032); Don't Screen Us Out (CGN0034); Royal College of Physicians of Edinburgh (CGN0036); Nuffield Council on Bioethics (CGN0049); Antenatal Results and Choices (CGN0075), para 3; Down's Syndrome Association (CGN0085)

237 Antenatal Results and Choices (CGN0075), para 3 and Don't Screen Us Out (CGN0034), para 1

help a prospective parent make an informed decision about the rest of their pregnancy".[238] Underscoring this, Dr Felicity Boardman of Warwick Medical School noted that there was a "significant contrast between the attitudes of people who live with genetic conditions and the general population on the possibility of selective reproduction", where "those with prior experience generally view the condition in a more favourable light than those without".[239]

74. Whereas submissions called for clear, balanced information for consumers using commercially-provided non-invasive prenatal testing, the Down's Syndrome Association told our predecessor Committee that there was "frequently an absence of support at the critical time of delivery of test results".[240] It added that "screening midwives, working within NHS hospitals, often speak in terms of being left to 'pick up the pieces' after women have paid privately for a commercial genomic testing service". The Nuffield Council on Bioethics recommended that "accurate, balanced and non-directive information should be readily available to women and couples in accessible written and multimedia formats", including "what they might expect from life with a child or adult with the condition being tested for".[241] Antenatal Results and Choices, a charity providing information and support to those undertaking antenatal screening, told our predecessor Committee that it "would be helpful if there could be easily accessible online guidance for both the providers and expectant parents considering such services, which is informed and endorsed by relevant international and national professional bodies and stakeholder organisations".[242]

75. Since our predecessor Committee decided to launch an inquiry into commercial genomics, there have been several significant developments regarding non-invasive prenatal testing. Firstly, the Care Quality Commission has started inspecting private clinics providing non-invasive prenatal testing.[243] Secondly, the Royal College of Obstetricians and Gynaecologists is developing guidance for healthcare professionals on care after non-invasive pre-natal testing, covering the provision of accurate testing and accurate, balanced and non-directive information and support, including how to deal with unanticipated or secondary findings and failed tests.[244] This is due to be published in "early 2021". The Nuffield Council on Bioethics has noted that this guidance could help to provide the Care Quality Commission's inspectors with professional standards specific to non-invasive prenatal testing to help them conduct their inspections.[245] The NHS has also updated its online information about Down's syndrome with help from the Down's Syndrome Association, with the Nuffield Council on Bioethics saying that the new website "now gives a much more balanced portrayal of Down's syndrome and what to expect from a life with the condition".[246]

238 Down's Syndrome Association (CGN0085)
239 Dr Felicity Boardman (CGN0012), section 3.3
240 Down's Syndrome Association (CGN0085)
241 Nuffield Council on Bioethics, 'Non-invasive prenatal testing: ethical issues' (2017), para 6.8 and Nuffield Council on Bioethics, 'Non-invasive prenatal testing: ethical issues—Short Guide' (2017), p14
242 Antenatal Results and Choices (CGN0075), para 6
243 Department of Health and Social Care and the Department for Business, Enterprise and Industrial Strategy (CGN0053), paras 65–67—see also: Nuffield Council on Bioethics, 'Care Quality Commission to regulate private providers of non-invasive prenatal testing following Nuffield Council recommendation', published 8 February 2019
244 Royal College of Obstetricians and Gynaecologists, 'Care after non-invasive pre-natal testing', published 31 October 2019
245 Nuffield Council on Bioethics, 'Non-invasive prenatal testing is starting to get the attention it deserves', published 23 September 2019
246 Nuffield Council on Bioethics, 'Non-invasive prenatal testing is starting to get the attention it deserves', published 23 September 2019

76. **The potential for results from non-invasive prenatal testing to influence decisions made on terminating pregnancies raises specific issues not encountered by most other genomic tests offered to consumers. Several submissions highlighted the importance of the information and other support provided to those receiving results from such tests to not only ensure comprehension of the result but also to provide balanced, non-directive information about the different options following the test result. Since our predecessor Committee launched its inquiry into direct-to-consumer genomics, there have been several significant developments related to the information and support provided with non-invasive prenatal testing. We hope these will address some of the concerns raised during our inquiry and our predecessor Committee's inquiry. *As the Government considers the requirements that should be introduced on the information provided to consumers using direct-to-consumers genomic tests, it should consider specific requirements for prenatal genomic testing to ensure that the information provided is balanced and non-directive, with accurate information on what might be expected from life for a child or adult with the condition being tested for.***

Support for the NHS

77. As described in paragraph 16, one concern regarding genomic tests provided directly to consumers was the risk that they could increase pressure on the NHS, in particular if consumers consulted NHS professionals to discuss test results that they had obtained privately. Although there is not yet strong evidence of this happening (see paragraphs 27 and 28), 23andMe recommends that its customers consult genetic counsellors before and after taking a test, and the then Government told our predecessor Committee that there were "scenarios in which consumers who have received a commercial genomic test result are likely to need or request access to counselling or support services in the NHS".[247] It said that "in this case, the consumer is most likely to approach their GP who can arrange referral to a specialist". It additionally warned that "any requirement to offer specialised NHS genetic counselling services to those receiving a commercial genomic test result would need to consider workforce capacity and potential new workforce models".[248]

78. The Royal College of General Practitioners has published a position statement advising general practitioners that "patients should not be referred to secondary or tertiary care solely on the basis of direct-to-consumer genomic test results", and that instead "patients should be offered the NHS care which would otherwise have been offered […] regardless of their direct-to-consumer genomic test result.[249] Noting that "NHS Genetics Centres have been advised not to see people if the test is not one that would be normally offered in the NHS", the Nuffield Council on Bioethics nevertheless warned that "clinicians have told us that it can be difficult to turn people away".[250] In 2019, the then Chair of the Royal College of General Practitioners told our predecessor Committee that general practitioners were "not sufficiently supported to interpret the results of direct-to-consumer genomic tests":

> As the availability of these tests to the wider public is a new phenomenon it is understandable that GPs and their teams may not have sufficient knowledge

247 23andMe (COG0002), para 4.6.1 and Department of Health and Social Care and the Department for Business, Enterprise and Industrial Strategy (CGN0053), para 36

248 Department of Health and Social Care and the Department for Business, Enterprise and Industrial Strategy (CGN0053), para 40

249 Royal College of General Practitioners and British Society for Genetic Medicine, 'Position Statement on Direct to Consumer Genomic Testing' (2019)

250 Nuffield Council on Bioethics (CGN0049), section 3

> and training of all areas around direct-to-consumer genomic testing to be able to interpret the results and appropriately support their patients. We believe it is likely that further education or training will be required for clinicians to help them understand the principles and terminology of genomics, as well as to understand the ethical issues that may arise; and to support patients, and implement appropriate management, and navigate new clinical pathways.[251]

The Royal College of General Practitioners said that it was working with NHS Health Education England to "develop a genomics toolkit for primary care professionals". The PHG Foundation suggested that "clear policies and guidance for NHS professionals on when and how commercially obtained testing results should be considered (and how to handle situations when they should not be) could help minimise the burden on NHS services considerably; such decisions require considerable thought and effective communication to the public".[252] The Royal College of General Practitioners added that it believed that the Government and NHS England should also "educate patients about the limitations of direct-to-consumer genomic testing".[253]

79. **Many submissions to this inquiry expressed concern that direct-to-consumer genomic testing could increase pressure on the NHS due to consumers consulting their GP following a test result. Although there is not yet strong evidence of this happening, some companies providing tests directly to consumers recommend that their customers consult genetic counsellors before and after using their products, and the previous Government acknowledged the likelihood of such customers consulting NHS specialists or GPs. *The Government should gather evidence on the current impact of direct-to-consumer genomic testing on the NHS, as well as the effectiveness of guidance and other support offered to NHS professionals encountering patients who have used such tests. If necessary, the Government should support the Royal Colleges and other relevant organisations to publish guidance for NHS professionals, as well as for consumers consulting the NHS following a direct-to-consumer genomic test, explaining the capabilities and limitations of those tests, how the NHS will act on results obtained from such tests and the reasons for the actions that the NHS will and will not take.***

80. The 2019 Topol Review of healthcare technology and its likely implications for NHS staff highlighted the growing role for genomics and the increasing need for NHS staff to be trained to work with genomic data.[254] Since 2014, NHS Health Education England has been running a Genomics Education Programme to develop genomics capability across the NHS workforce.[255] In 2019, the Association of Genetics Nurses and Counsellors warned, however, that its workforce in particular was in a "dire situation":

> The potential risks from an increase in the availability of commercial genomic testing is that it increases burden on an already stretched NHS

251 Letter from Prof Helen Stokes-Lampard to Rt Hon Sir Norman Lamb MP, 7 October 2019
252 PHG Foundation (CGN0023), para 38—see also: The Association of the British Pharmaceutical Industry (CGN0066), para 16
253 Letter from Prof Helen Stokes-Lampard to Rt Hon Sir Norman Lamb MP, 7 October 2019
254 NHS, 'The Topol Review: Preparing the healthcare workforce to deliver the digital future' (2019)
255 NHS Health Education England, 'Genomics Education Programme: About Us', accessed 12 May 2021

> genetic counselling service. In the UK there are only 300 genetic counselling positions and due to retirement and unfilled posts we will be short of 80 genetic counsellors in the next 3 years.[256]

Professor Mark Caulfield, Chief Scientist at Genomics England, suggested that the "use of automated counselling […] may help to address the workforce capacity to deal with [increased demand for counselling]".[257] However, as discussed at paragraph 63, Professor Anna Middleton, Chair of the Association of Genetics Nurses and Counsellors, argued that meaningful counselling could not be delivered using automated systems.[258]

81. The Royal Colleges of Physicians and of Pathologists raised concern that the current situation, where consumers may look to the NHS to interpret results obtained from direct-to-consumer tests, created "an uneven playing-field where the commercial providers take profit from offering tests whilst contributing nothing to the NHS providers who are left dealing with unexpected or difficult outcomes".[259] The Cancer Genetics Group recommended that the Government should "consider a private company levy to help pay for NHS validation and interpretation of findings".[260] Asked by our predecessor Committee about the industry contributing to the cost of training genetic counsellors, representatives from 23andMe, Ancestry and DNAfit indicated that their companies would be willing to support such an initiative.[261] In a letter responding to the previous Committee's interim findings, the then Minister, Baroness Blackwood, said that Health Education England would be willing to "explore [with stakeholders in the commercial genomics sector] how they may support an increase in training commissions for genetic counsellors".[262] The current Minister, Lord Bethell, told us that the companies' offer was "very welcome" and that Health Education England was in the "early stages of working with NHS England and system partners to assess how it can collaborate with the private sector on genetic counsellor training".[263]

82. **Concern was raised with us that companies providing genomic tests directly to consumers could profit from supplying the tests while leaving the NHS to deal with consumers and their results following the test. Addressing this issue, representatives of several major direct-to-consumer testing companies indicated to our predecessor Committee their willingness to contribute to ongoing efforts to train genetic counsellors within the NHS. *The Government should continue to explore, with NHS England and NHS Health Education England, the opportunity for companies selling genomic tests directly to consumers to contribute to the costs of training genetic counsellors in the NHS.***

Data protection

83. As discussed in paragraphs 21–22, several concerns were raised regarding the privacy and consent of consumers providing samples to companies for genomic sequencing, although few of these concerns were specific to genomic testing. Genetic data processed in

256 Association of Genetic Nurses and Counsellors (CGN0008)
257 Q59
258 Oral evidence taken on 15 October 2019, HC (2019) 33, Q29
259 The Royal College of Physicians and the Royal College of Pathologists (CGN0022)
260 Cancer Genetics Group (CGN0007), table 1
261 Oral evidence taken on 28 October 2019, HC (2019) 33, Q130
262 Letter from Baroness Blackwood to the Chair of the Science and Technology Committee, 28 January 2020
263 Lord Bethell, Parliamentary Under Secretary of State for Innovation (COG0009)

the UK is subject to data protection legislation, under which it is given the same protection as certain other "special categories of personal information" (such as data revealing ethnic origin or religious beliefs).[264] Consequently, genetic data can only be processed if the relevant individual has given explicit consent for their data to be processed for a specified purpose, other than in certain other circumstances (for example for reasons of public interest, such as for public health). This may not apply, however, where a consumer transfers their data out of the UK for processing, or their genetic sample out of the UK for sequencing. Representatives of 23andMe, Ancestry and DNAfit each told our predecessor Committee that their companies complied with data protection legislation, and that where they shared consumer data it was always with the consumers' continued consent and in a de-identified form.[265] Although EthicsAndGenetics, a campaign group, argued that the "business model of companies such as 23andMe and Ancestory.com is to harvest genomic and health data from the people that purchase their genetic tests, and sell that data for their own profit", Kathy Hibbs, Chief Legal and Regulatory Officer for 23andMe, said that 23andMe "generate[d] more income from customers than we do from our data-sharing arrangements".[266]

84. With regards to specific concerns about the potential use of genetic data by insurers, a Code on Genetic Testing and Insurance has been agreed between the Government and British insurers that permits insurers to consider the results of a predictive genetic test only for applications for life insurance above £500,000, and where the test result relates to Huntington's disease.[267]

85. Despite this legislation and the Code on Genetic Testing, several submissions argued that problems persisted.[268] For example, researchers at Newcastle University told our predecessor Committee that "online formats easily conflate informed consent with current practices around tick-boxing terms and conditions", and that "the provider has no idea if the individual has even read the contract, never mind understood it".[269] Ms Hibbs, however, highlighted that the consent processes used by 23andMe were approved by an external ethics board, to ensure that "they give people the right amount of information to make it clear what they are consenting to and what the risks are":

> [Consumers] have to proactively consent and then, presuming that they have consented, any time they are in their account it says at the top, 'You're currently consented to research. Click here if you want to change that consent'. It is very easy to remind them that they are consented and very easy for them to change.[270]

Carla Newell, Chief Legal Officer and Chief Risk Officer at Ancestry, said that its data-sharing collaborations were also reviewed and transparently communicated on the

264 See: European Union (Withdrawal) Act 2018, section 3; Data Protection, Privacy and Electronic Communications (Amendments etc) (EU Exit) Regulations 2019 (SI 2019/419); Regulation 2016/679 of the European Parliament and of the Council, Articles 4 and 9; Data Protection Act 2018—this does not apply if the individual in question is not identifiable from the data held.

265 Oral evidence taken on 28 October 2019, HC (2019) 33, Qq131–144

266 EthicsAndGenetics (CGN0042), para 6 and oral evidence taken on 28 October 2019, HC (2019) 33, Q144

267 HM Government and Association of British Insurers, 'Code on Genetic Testing and Insurance' (2018)

268 For example, see: The Royal Society (CGN0019); Nuffield Council on Bioethics (CGN0049), section 4; Dr Pauline McCormack *et al.* (CGN0057), section 7b

269 Dr Pauline McCormack *et al.* (CGN0057), section 7b

270 Oral evidence taken on 28 October 2019, HC (2019) 33, Qq137–138

company's website.[271] Several submissions nevertheless emphasised the difficulty for consumers to know how their data was being used and what risks that might entail.[272] The British Pharmacological Society suggested that the "complexity of ethical, legal, and social issues surrounding consent for genomic testing indicate that substantial effort is required to ensure adequate understanding of the test by consumers", and that "depending on how many of these issues apply, professional genetic counselling may be crucial for obtaining truly informed consent for genomic tests".[273] Dr Andelka Phillips, of the University of Waikato, recommended that regulators publish specific public guidance for direct-to-consumer genomic testing to explain how generic regulation such as the General Data Protection Regulation applied to such testing, referencing the Office of the Canadian Privacy Commissioner's policy statements as an example.[274] She argued that this could "provide the public with access to more independent informational resources to assist them in making informed decisions about whether or not to utilise commercial genomics services". Regarding the use of genomic data for criminal justice, Dr Matthew Hurles, Head of Human Genetics at the Wellcome Sanger Institute, said that "having clarity from the Government about what the Home Office can and cannot do would, again, help to build trust" with those sharing their data.[275]

86. With regards to the relevance of an individual's genomic data for their relatives—one of the main reported differences between genomic data and other forms of personal data when considering privacy and consent—Dr Felicity Boardman, Associate Professor in Medicine, Ethics and Society at Warwick Medical School, wrote that it raised questions about a possible "obligation of disclosure to these relatives" as well as, conversely, the "question of whether biological relatives of [consumers purchasing tests] have the right not to know information about their genetic health".[276] Professor Anneke Lucassen, then Chair of the British Society for Genetic Medicine, agreed that "we need to think more widely about how we let [affected relatives] know, and how we let them know respecting their possible wishes not to know".[277] The Shelford Group, a collaboration between ten of the largest research and teaching NHS trusts in England, suggested that "an individual receiving a genomic test should be aware of their relatives' wishes prior to testing".[278] A recent court case established the legal responsibilities of healthcare professionals to disclose genetic results to potentially affected family members,[279] but it is unclear what responsibilities apply or should apply to other individuals or companies.

87. The Royal Society noted that "as data enhances our analytical capabilities, notions such as consent, privacy and ownership are becoming more difficult to maintain", and that "as a result, many of the concepts that sit at the core of public confidence in governance

271 Oral evidence taken on 28 October 2019, HC (2019) 33, Qq142–143

272 For example, see: Dr Felicity Boardman (CGN0012), section 3.1; Dr Elizabeth Ormondroyd (CGN0061), section 5

273 British Pharmacological Society (CGN0027), para 7.2—see also: Dr Pauline McCormack *et al.* (CGN0057), section 7b

274 Dr Andelka Phillips (CGN0025)—see also: 'Direct-to-consumer genetic testing and privacy', Office of the Canadian Privacy Commissioner, accessed 25 May 2021

275 Oral evidence taken on 15 October 2019, HC (2019) 33, Q52

276 Dr Felicity Boardman (CGN0012), section 3.2—see also: The Royal Society (CGN0019), para 4; British Pharmacological Society (CGN0027), para 7.1; Association of Medical Research Charities (CGN0028), para 20; Shelford Group (CGN0037), para 19

277 Oral evidence taken on 15 October 2019, HC (2019) 33, Q46

278 Shelford Group (CGN0037), para 19

279 ABC vs St George's Healthcare NHS Trust, South West London and St George's Mental Health NHS Trust and Sussex Partnership NHS Foundation Trust, [2020] EWHC 455 (QB)—see also: NHS Resolution, 'Case of note: ABC v St George's Healthcare NHS Trust and Others (Court of Appeal, 16 May 2017)', published 17 August 2017; PHG Foundation, 'After ABC v St George's: a new duty to consider', published 3 March 2020

are no longer fit for purpose".[280] As one example, it said that the "ability to draw connections between data is now so advanced that approaches to managing privacy, such as deidentification, may no longer apply".[281] The Cancer Genetics Group added that "as the predictive power of genomic data and linked data sets improve, insurance companies may start to become increasingly concerned about premiums".[282] The Nuffield Council on Bioethics argued that technological advances could also, however, help to resolve challenges associated with privacy and consent, suggesting that blockchain technologies could give individuals greater control over the uses of their data.[283]

88. **Various concerns related to privacy and consent regarding data generated by direct-to-consumer genomic tests were raised during this inquiry. Many of these were similar to concerns that have been expressed regarding personal data more generally, although the relevance of an individual's genomic data to that individual's relatives was raised as a particular feature of genomic data. Despite data protection legislation and voluntary agreements such as the Code on Genetic Testing and Insurance, the complexity and uncertainty of future uses of genomic data may challenge current procedures for obtaining informed consent. Further, consumers may not benefit from this data protection if they transfer their data or genetic samples out of the UK for processing. As technologies develop and more consumers use direct-to-consumer genomic tests, existing data safeguards may become less effective and the consequences for privacy more significant. *The Government should aim for the data protection framework governing genomic data in the UK to be world-leading. It should review the adequacy of the UK's data protection framework for direct-to-consumer genomic testing, including the risks and opportunities presented by technological developments and growing numbers of consumers using direct-to-consumer genomic tests. The Government should also consider the case for requiring companies providing direct-to-consumer genomic tests to inform consumers, at the point of sale, of the potential consequences of genomic test results for their relatives.***

Permitted uses of genomic testing

89. Researchers at Newcastle University suggested to our predecessor Committee that any genomic test worth having would be available to appropriate individuals on the NHS, and that there was therefore no need for health-related genomic testing to be available for purchase by consumers.[284] However, Dr Matthew Hurles, Head of Human Genetics at the Wellcome Sanger Institute, and Professor Anneke Lucassen, then Chair of the British Society for Genetic Medicine, acknowledged that there were certain instances in which genomic testing could have clinical benefit but was not offered by the NHS (such as pharmacogenetic testing, which can help determine the most appropriate treatment for an individual).[285] For example, Dr Hurles pointed out that the NHS "tries to come to a balance as regards cost-effectiveness", which could restrict its use of expensive tests with limited

280 The Royal Society (CGN0019), para 4
281 The Royal Society (CGN0019), para 4
282 Cancer Genetics Group (CGN0007), para 12
283 Nuffield Council on Bioethics (CGN0049), section 4
284 Dr Pauline McCormack *et al.* (CGN0057), section 1
285 Oral evidence taken on 15 October 2019, HC (2019) 33, Qq10–12 and 54—these included epigenetic testing, prenatal testing for medium-risk pregnancies, pre-conception testing and pharmacogenetic testing. Several commercial providers of genomic testing further argued that there were uses for such testing in a wider range of applications not yet used by the NHS, such as to guide screening for certain diseases—see: Everything Genetic Ltd (CGN0005); Congenica Limited (CGN0046), section 4a; Genomics plc (CGN0048), paras 23–31

but nevertheless real benefit.[286] Dr Ron Zimmern, Chair of the PHG Foundation, argued that, whereas the "NHS will necessarily need to be more cautious and more risk averse [than commercial actors]", to "use that as an excuse to prohibit a more entrepreneurial private sector to implement services that might benefit the population would be misguided and slow down the development of services that might be beneficial".[287]

90. Some, such as the Association of Medical Research Charities, questioned the ethics of allowing people to receive results that could cause upset.[288] Dante Labs, however, said that "DNA belongs to the individuals" and that "people have the right to learn about their DNA and have the right to choose where to get their DNA analysed".[289] The Minister for Innovation, Lord Bethell, argued that the Government "cannot expect people somehow to be protected from their own genetic data":

> My instincts are that we should not put people in cotton wool on this issue. We allow people to buy their own pensions, get divorced and take exams, the results of which are posted to them in envelopes that are traumatic to open.[290]

Highlighting particular issues with the genomic testing of children, researchers at Newcastle University that they "would endorse differential regulation for tests with different purposes, with an 'in principle' restriction of some tests" being provided directly to consumers.[291] Specific examples are discussed below.

Testing children

91. The issue of direct-to-consumer genomic testing on asymptomatic children for conditions that onset in adulthood, and for which no treatments or preventative interventions are available during childhood, was raised in paragraph 23. A 2015 survey of a representative sample of the British population found that more people thought that such testing should be permissible (47%) than should not be (20%).[292] However, a range of professional bodies in the UK and internationally, as well as the Human Genetics Commission (a former advisory body to the UK Government), have recommended that such testing should not normally be provided directly to consumers.[293] Despite noting some potential benefits of testing for adult-onset conditions in childhood, the European Society of Human Genetics argued, for example, that unless preventative measures were available during childhood, such testing "should be deferred until the person has the maturity and competence to understand the nature of the decision and its implications":

286 Oral evidence taken on 15 October 2019, HC (2019) 33, Q10
287 Dr Ron Zimmern (CGN0020), para 28
288 Association of Medical Research Charities (CGN0028), para 22—see also: Macmillan Cancer Support (CGN0051)
289 Dante Labs SRL (CGN0018)
290 Q55
291 Dr Pauline McCormack et al. (CGN0057), section 7
292 Shkedi-Rafid *et al.*, 'Genetic testing of children for adult-onset conditions: opinions of the British adult population and implications for clinical practice', European Journal of Human Genetics, vol 23 (2015)
293 British Society for Human Genetics, 'Report on the Genetic Testing of Children' (2010); American College of Medical Genetics and Genomics, 'Technical report: ethical and policy issues in genetic testing and screening of children', Genetics in Medicine, vol 15 (2013); European Society of Human Genetics, 'Statement of the ESHG on direct-to-consumer genetic testing for health-related purposes', European Journal of Human Genetics, vol 18 (2010) and Human Genetics Commission, 'A Common Framework of Principles for direct-to-consumer genetic testing services' (2010), principle 6.9

> The risk to relatives, the absence of an effective cure, the potential loss of health insurance, the financial costs of testing and the inability to 'undo' the knowledge have been identified as reasons why adults decide not to undergo predictive genetic tests for adult-onset disorders. Considering that minors, far more than their parents, will be living with the repercussions of the test results, there are good reasons that they should be able to decide about the participation in such a genetic test.[294]

92. The American College of Medical Genetics and Genomics has noted the "wide variation in preferences among adults" with regards to deciding whether or not to obtain a genomic test, and observed that "commentators argue that allowing parents or guardians to test their minor children unfairly pre-empts the future choices of those children" and recommended that genetic testing for late-onset conditions be deferred until adulthood.[295] Professor Anneke Lucassen, then Chair of the British Society of Genetic Medicine, observed that "most" adults who initially wanted to have a test for Huntington's disease (an untreatable genetic condition that manifests in later life) from the NHS ultimately decided against the test once they had had the opportunity to talk it through with a clinical genetics professional.[296]

93. Genomic testing on children for non-medical purposes, such as aptitude for sport or music, was also referenced in the written evidence (see paragraph 23).[297] Researchers at Newcastle University stated that they saw "no benefits in genomic testing of children outside of a healthcare context".[298] Dr Andelka Phillips of the University of Oxford warned of the "dubious validity" of "child talent tests" and argued that they should be banned and that regulators should alert the public about the most problematic services.[299]

94. Currently, there is no legislation preventing parents from obtaining genomic tests for their children. Some of the largest direct-to-consumer testing companies permit children to take a test provided their parents or guardians consent.[300] Researchers from Newcastle University told our predecessor Committee that a "number of direct-to-consumer genomic testing companies specifically encourage the testing of children".[301] Consequently, some submissions called for the rules on testing asymptomatic children to be reviewed. The Shelford Group of NHS Trusts recommended that "asymptomatic children should not be tested" using any genomic tests and DNAfit, a company that sells genomic tests directly to consumers, said that companies should obey the principle of "no direct-to-consumer genetic testing for under-18s".[302] The Nuffield Council on Bioethics recommended that companies should not carry out genomic tests on children that do not meet the criteria

294 European Society of Human Genetics, 'Genetic testing in asymptomatic minors: recommendations of the European Society of Human Genetics', European Journal of Human Genetics, vol 17 (2009) and European Society of Human Genetics, 'Genetic testing in asymptomatic minors: Background considerations towards ESHG Recommendations', European Journal of Human Genetics, vol 17 (2009)

295 American College of Medical Genetics and Genomics, 'Technical report: ethical and policy issues in genetic testing and screening of children', Genetics in Medicine, vol 15 (2013)

296 Oral evidence taken on 15 October 2019, HC (2019) 33, Q30

297 For example, see: Dr Andelka Phillips (CGN0025); Nuffield Council on Bioethics (CGN0049), section 5; Dr Pauline McCormack *et al.* (CGN0057), section 7b; Dr Peter Fotheringham (CGN0082), paras 1–2, 7 and 14–16

298 Dr Pauline McCormack *et al.* (CGN0057), section 7b

299 Dr Andelka Phillips (CGN0025)

300 Ancestry.com, 'Activating an AncestryDNA® Kit for Your Child'; MyHeritage, 'Terms and Conditions' and Family Tree DNA, 'Terms of Service', all accessed 11 January 2021

301 Dr Pauline McCormack et al. (CGN0057), section 7b

302 Shelford Group (CGN0037), para 8 and Prenetics International and DNAfit (CGN0035), section 7

of the UK National Screening Committee.[303] Among other things, application of these criteria would prevent testing of asymptomatic children for susceptibility to non-serious conditions or those where no effective treatment or intervention is known (such as for Alzheimer's disease).[304]

95. **Although there is potential for the results of a genomic test to be upsetting, this is not a sufficient reason to prevent consenting adults from using these tests. However, a range of submissions to our inquiry and our predecessor Committee's inquiry highlighted a potential need for restrictions on direct-to-consumer genomic testing of children. Professional bodies in the UK and internationally have recommended that genomic tests are not provided directly to consumers for the testing of asymptomatic children for adult-onset conditions for which no intervention can be made during childhood. *The Government should consider which, if any, genomic tests for asymptomatic children should be able to be provided directly to consumers, including whether there should be a ban on the provision of genomic tests for use on children that do not meet the criteria of the UK National Screening Committee.***

Prenatal testing

96. A related class of genomic testing is prenatal testing. Although many of the issues related to prenatal testing are similar to any other form of genomic testing, the 'moral status' of a fetus makes termination of a fetus an option that is not applicable for children or adult users. Additionally, diseases may be apparent in a child but not in a fetus, making asymptomatic testing relevant to a wider range of conditions. In particular, the Nuffield Council on Bioethics flagged non-invasive prenatal testing to our predecessor Committee, which is a technique that uses placental DNA circulating in the mother's blood to obtain genetic data from the fetus without the invasive procedures required for traditional techniques, and the commercial use of which has which has been growing since 2011.[305]

97. As described in paragraphs 14 and 15, prenatal genomic testing can provide benefits to prospective parents but has also raised concerns related to its ability to influence decisions on terminations of fetuses. The Nuffield Council on Bioethics reported that "research on the views of pregnant women and families with direct experience of genetic conditions has found widespread support for the availability of non-invasive prenatal testing to detect rare genetic conditions".[306] Despite the concerns expressed by some submissions, none explicitly advocated for prenatal testing to be completely banned for consumers.

98. Instead of a ban on all prenatal genomic testing available to consumers, the Nuffield Council on Bioethics recommended that non-invasive prenatal genomic testing "should only be offered for significant medical conditions or impairments that manifest in childhood", and "normally should not be used to test whether:

- a fetus has a less significant medical condition, or an adult onset condition;

303 Nuffield Council on Bioethics (CGN0049), section 5—see also: Nuffield Council on Bioethics, 'Medical profiling and online medicine: the ethics of 'personalised healthcare' in a consumer age' (2010), para 9.42

304 Public Health England, 'Criteria for appraising the viability, effectiveness and appropriateness of a screening programme', accessed 8 January 2021

305 Nuffield Council on Bioethics (CGN0049), section 1—see also: Nuffield Council on Bioethics, 'Non-invasive prenatal testing: ethical issues' (2017), paras 1.1–1.4; M. Minear *et al.*, 'Global perspectives on clinical adoption of NIPT', Prenatal Diagnosis, vol 35 (2015); IMARC, 'Non-Invasive Prenatal Testing Market: Global Industry Trends, Share, Size, Growth, Opportunity and Forecast 2021–2026' (2020)

306 Nuffield Council on Bioethics, 'Non-invasive prenatal testing: ethical issues' (2017), para 1.27

- 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)".[307]

Explaining the rationale for this recommendation, the Council said that, in addition to concerns of increased rates of terminations and of removing individuals' rights to making decisions for themselves as adults:

> A further concern is that, given that [the information generated by prenatal testing] usually would not be grounds for termination and would have no clinical use prenatally, offering such tests could be regarded as not meeting the responsibilities of health and social professionals to ensure that all patients receive good care and treatment.[308]

The Nuffield Council on Bioethics conceded, however, that it was "possible to imagine several exceptions to [its] recommendation".[309] It gave the specific example of a woman or a couple with a family history of an adult onset condition wanting to find out if their fetus would develop the condition, if the condition was extremely serious and manifested in mid-life, if there was no treatment available, and if termination of pregnancy was an option (these criteria are met by Huntington's disease, testing for which is already available). It argued that the fact that tests were already available should not always provide grounds for the continued provision of testing though, noting that non-invasive prenatal testing to determine sex was already "widely available in the UK through private companies" but recommending that the Government "should require test providers to neither generate nor report [sex determination] information unless there is concern that the fetus may be showing signs of a significant sex chromosome aneuploidy or is at risk of a sex-linked disorder" (on the basis that this could increase the risk of terminations taking place based on the sex of the fetus).[310]

99. **Prenatal genomic testing could influence decisions on terminating fetuses, leading to specific concerns in addition to the issues concerning other types of direct-to-consumer genomic testing.** ***The Government should consider if any restrictions should be placed on the conditions that prenatal genomic tests provided directly to consumers are able to test for.***

Regulatory scope

Regulating an international market

100. The PHG Foundation noted that, often, "consumers access test kits via the Internet and access results by email or via a secure site".[311] The Nuffield Council on Bioethics warned that this "international nature of the market raises challenges for policy makers in the UK".[312] Indeed, Graeme Tunbridge, Director of Devices at the Medicines and

307 Nuffield Council on Bioethics (CGN0049), section 1—see also: Nuffield Council on Bioethics, 'Non-invasive prenatal testing: ethical issues' (2017), paras 6.4 and 6.12–6.17
308 Nuffield Council on Bioethics, 'Non-invasive prenatal testing: ethical issues' (2017), para 6.13
309 Nuffield Council on Bioethics, 'Non-invasive prenatal testing: ethical issues' (2017), para 6.14
310 Nuffield Council on Bioethics, 'Non-invasive prenatal testing: ethical issues' (2017), paras 6.15–16
311 PHG Foundation (CGN0023), para 36
312 Nuffield Council on Bioethics (CGN0049), section 5

Healthcare products Regulatory Agency, told us that there was "relatively little in the way of regulation where the test is provided outside the EU".[313] The then Government clarified to our predecessor Committee that this was the case where the genomic sequencing was conducted outside of the European Union, even if the consumer purchased the test and took the sample within it.[314]

101. ***The Government should consider requiring any manufacturer making genomic tests available to consumers in the UK to register a legal representative in the UK, with responsibility for ensuring that products supplied to consumers in the UK meet all relevant UK regulatory requirements.***

Secondary analysis

102. The Nuffield Council on Bioethics noted that "some companies offer to provide people with raw sequencing data alongside their interpretation of the results", which allows the consumer to "go to other companies for further interpretation services".[315] Indeed, a 2018 study found that around 67% of consumers who had used a direct-to-consumer genomic test had used a third-party service to re-interpret their sequence data.[316] Professor Anneke Lucassen, then Chair of the British Society for Genetic Medicine, expressed her "criticisms" of these secondary analysis services in particular.[317] The British Society for Genetic Society explained that consumers could, for example, obtain data from a test intended for ancestry purposes and ask a second company to analyse it for medically-related purposes, despite the unsuitability of the test technology and methodology for that purpose.[318]

103. Genomic tests sold directly to consumers for medically-related purposes are currently regulated as an 'in vitro diagnostic medical device', which the regulations specify means a "medical device which is a reagent, reagent product, calibrator, control material, kit, instrument, apparatus, equipment or system, whether used alone or in combination" (intended to be used for a variety of specific purposes).[319] The then Government told our predecessor Committee that these regulations "do not cover the regulation of testing services".[320] The regulations that would have been introduced under the previous Government would have expanded the scope of products covered, explicitly adding "software" to the list of what could constitute an in vitro diagnostic medical device.[321] The PHG Foundation said that this could provide "another potential means of oversight".[322]

313 Q25
314 Department of Health and Social Care and the Department for Business, Enterprise and Industrial Strategy (CGN0053), para 63
315 Nuffield Council on Bioethics (CGN0049), section 2
316 Wang *et al.*, 'Consumer use and response to online third-party raw DNA interpretation services', Molecular Genetics and Genomic Medicine, vol 6 (2018)
317 Oral evidence taken on 15 October 2019, HC (2019) 33, Q15
318 British Society for Genetic Medicine (CGN0030), section 4—see also oral evidence taken on 28 October 2019, HC (2019) 33, Qq79–83
319 The Medical Devices Regulations 2002 (SI 2002/618), regulation 2
320 Department of Health and Social Care and the Department for Business, Enterprise and Industrial Strategy (CGN0053), para 55
321 The Medical Devices (Amendment etc.) (EU Exit) Regulations 2019 (SI 2019/791), regulation 11—see also: Directive 98/79/EC of the European Parliament and of the Council, Article 1; Regulation 2017/746 of the European Parliament and of the Council, Article 2
322 PHG Foundation (CGN0023), para 32

104. **As well as companies offering products to consumers that combine genomic testing with analysis of the genomic data obtained through the test, some companies offer secondary analysis of genomic data obtained through a previous genomic test from a different company. There is a potentially increased risk that the testing process used to obtain the genomic data is unsuitable for the analysis performed by a company offering secondary analysis. Despite this, the current regulation may not apply to companies providing secondary analysis of genomic data if their product is not deemed to include the use of physical equipment.** ***The Government should consider extending the definition of products covered by the regulation of genomic tests to include software and other services offering analysis and interpretation of genomic test results obtained from third parties.***

Conclusions and recommendations

The benefits and risks of direct-to-consumer genomic testing

1. A range of benefits and concerns have been raised regarding the availability of genomic testing for direct purchase and use by consumers. These apply to all types of genomic tests, but are arguably most acute for tests used for medically-related purposes. Despite concerns around direct-to-consumer tests existing for many years, evidence of harm has mostly not been systematically collected and is limited. As direct-to-consumer genomic testing becomes more widespread and covers a greater variety of conditions, evidence of positive impacts and harms may grow. We have heard calls for updated regulation of direct-to-consumer genomic testing. While the Government has acknowledged the importance of maintaining public trust in genomics and said that it would "establish a gold standard UK model for how to apply strong and consistent ethical and regulatory standards", it has committed only to consulting on and publishing future regulations for Great Britain at a "later date". *The Government should set out a specific timeframe in which it intends to review the case for introducing new regulations for genomic tests provided directly to consumers ('direct-to-consumer genomic tests').* (Paragraph 34)

Supporting a responsible direct-to-consumer genomic testing industry

2. Strong support for genomic testing from the current and previous Governments has helped to make the UK a world-leader in the sector. Although this support has focused on testing in the NHS, it has nonetheless supported the direct-to-consumer genomic testing industry. *The Government should continue its support for genomic testing in the UK.* (Paragraph 37)

3. Most manufacturers of genomic tests sold directly to consumers can self-certify the conformity of their products to performance requirements. The Medicines and Healthcare products Regulatory Agency has suggested that this restricts its ability to ensure that genomic tests on the UK market provide reliable results. *The Government should require manufacturers of direct-to-consumer genomic tests to have the performance of their tests assessed by an external body prior to placing their products on the UK market.* (Paragraph 41)

4. The results obtained from genomic tests provided directly to consumers cannot be integrated into Genomics England or NHS records since they do not always meet the standards required of the end-to-end testing and data handling process. Enabling tests that are provided directly to consumers to demonstrate that they meet the required standards could help to: reduce the likelihood of false positive or false negative results; facilitate the sharing of data obtained from direct-to-consumer tests, which could potentially support research efforts; and reduce the need for the NHS to re-test individuals following a commercially-obtained test, potentially reducing the burden placed on the NHS by direct-to-consumer tests. *The Government should work with Genomics England and the NHS to define clear technical standards for direct-to-consumer genomic testing that, if met, would enable the genomic data generated by the test to be used and trusted by Genomics England*

and the NHS. The Government should also establish a mechanism by which providers of direct-to-consumer genomic tests could validate that their tests met these standards. (Paragraph 44)

5. The performance requirements on direct-to-consumer genomic tests under the current regulations focus on a genomic test's analytical performance, not its clinical performance. For a medically-relevant test, however, clinical performance is fundamental to how the test will be used by a consumer. *The Government should extend the scope of the performance requirements on direct-to-consumer genomic tests to explicitly cover clinical performance as well as analytical performance.* (Paragraph 47)

6. As the evidence base for genomic testing develops rapidly, some have expressed criticism of the evidence used by some direct-to-consumer genomic testing companies to justify their tests, or warned of the risk of companies 'cherry picking' the most favourable evidence available. Requiring external validation of direct-to-consumer genomic tests, covering clinical as well as analytical performance, could help to address this. *In addition to pre-market validation of direct-to-consumer tests, the Government should consider requiring companies offering such tests to regularly update the evidence submitted to the external validation body, and for that body to review this, for example on an annual basis.* (Paragraph 53)

7. Results obtained from genomic testing must typically be considered in the context of an individual's specific circumstance—including their symptoms, personal and family medical history and ethnicity—in order for the clinical significance of those results to be interpreted correctly. These personal details are also often pertinent to the selection of the most appropriate genomic test for an individual. The NHS takes account of these contextual factors through the expert supervision and counselling provided throughout the testing process. In contrast, companies providing genomic testing directly to consumers appear to provide generic tests and depend upon consumers understanding written information provided alongside the test that has not been tailored to their personal situation. *The Government should consider the case for amending the regulation of genomic tests provided directly to consumers, to require medical supervision or the provision of genetic counselling for at least some types of genomic testing offered directly to consumers. Criteria used to determine which tests should require medical supervision could include the severity of the conditions being tested for, as well as the predictive power of the test. Requirements for supervision and genetic counselling should cover the qualifications of the medical intermediary required and minimum requirements on the content and format of the support or oversight provided.* (Paragraph 64)

8. Several contributors to this inquiry expressed concern that the information provided to consumers before and after using a direct-to-consumer genomic test, as well as the advertising used to market direct-to-consumer genomic tests, did not do enough to address public misconceptions of the capability of these tests and clarify the clinical utility of the results generated. Even where advertising material has used statistics accurately, the Advertising Standards Authority has ruled that there is still scope for them to provide a misleading impression to consumers. *The Government should consider the case for including reviews of the information provided to consumers*

prior to and after taking a direct-to-consumer test within any external validation required to place such tests on the market. This could, for example, include assessment of studies of consumer understanding of the information provided. (Paragraph 71)

9. *Building on its review of advertising for non-invasive prenatal testing, the Advertising Standards Authority should review, within the next year, the marketing materials used by companies offering other genomic tests directly to consumers, focusing in particular on the clinical performance implied by the tests compared with their actual performance.* (Paragraph 72)

10. The potential for results from non-invasive prenatal testing to influence decisions made on terminating pregnancies raises specific issues not encountered by most other genomic tests offered to consumers. Several submissions highlighted the importance of the information and other support provided to those receiving results from such tests to not only ensure comprehension of the result but also to provide balanced, non-directive information about the different options following the test result. Since our predecessor Committee launched its inquiry into direct-to-consumer genomics, there have been several significant developments related to the information and support provided with non-invasive prenatal testing. We hope these will address some of the concerns raised during our inquiry and our predecessor Committee's inquiry. *As the Government considers the requirements that should be introduced on the information provided to consumers using direct-to-consumers genomic tests, it should consider specific requirements for prenatal genomic testing to ensure that the information provided is balanced and non-directive, with accurate information on what might be expected from life for a child or adult with the condition being tested for.* (Paragraph 76)

11. Many submissions to this inquiry expressed concern that direct-to-consumer genomic testing could increase pressure on the NHS due to consumers consulting their GP following a test result. Although there is not yet strong evidence of this happening, some companies providing tests directly to consumers recommend that their customers consult genetic counsellors before and after using their products, and the previous Government acknowledged the likelihood of such customers consulting NHS specialists or GPs. *The Government should gather evidence on the current impact of direct-to-consumer genomic testing on the NHS, as well as the effectiveness of guidance and other support offered to NHS professionals encountering patients who have used such tests. If necessary, the Government should support the Royal Colleges and other relevant organisations to publish guidance for NHS professionals, as well as for consumers consulting the NHS following a direct-to-consumer genomic test, explaining the capabilities and limitations of those tests, how the NHS will act on results obtained from such tests and the reasons for the actions that the NHS will and will not take.* (Paragraph 79)

12. Concern was raised with us that companies providing genomic tests directly to consumers could profit from supplying the tests while leaving the NHS to deal with consumers and their results following the test. Addressing this issue, representatives of several major direct-to-consumer testing companies indicated to our predecessor Committee their willingness to contribute to ongoing efforts to train genetic counsellors within the NHS. *The Government should continue to explore, with*

NHS England and NHS Health Education England, the opportunity for companies selling genomic tests directly to consumers to contribute to the costs of training genetic counsellors in the NHS. (Paragraph 82)

13. Various concerns related to privacy and consent regarding data generated by direct-to-consumer genomic tests were raised during this inquiry. Many of these were similar to concerns that have been expressed regarding personal data more generally, although the relevance of an individual's genomic data to that individual's relatives was raised as a particular feature of genomic data. Despite data protection legislation and voluntary agreements such as the Code on Genetic Testing and Insurance, the complexity and uncertainty of future uses of genomic data may challenge current procedures for obtaining informed consent. Further, consumers may not benefit from this data protection if they transfer their data or genetic samples out of the UK for processing. As technologies develop and more consumers use direct-to-consumer genomic tests, existing data safeguards may become less effective and the consequences for privacy more significant. *The Government should aim for the data protection framework governing genomic data in the UK to be world-leading. It should review the adequacy of the UK's data protection framework for direct-to-consumer genomic testing, including the risks and opportunities presented by technological developments and growing numbers of consumers using direct-to-consumer genomic tests. The Government should aim for the data protection framework governing genomic data in the UK to be world-leading. It should review the adequacy of the UK's data protection framework for direct-to-consumer genomic testing, including the risks and opportunities presented by technological developments and growing numbers of consumers using direct-to-consumer genomic tests. The Government should also consider the case for requiring companies providing direct-to-consumer genomic tests to inform consumers, at the point of sale, of the potential consequences of genomic test results for their relatives.* (Paragraph 88)

14. Although there is potential for the results of a genomic test to be upsetting, this is not a sufficient reason to prevent consenting adults from using these tests. However, a range of submissions to our inquiry and our predecessor Committee's inquiry highlighted a potential need for restrictions on direct-to-consumer genomic testing of children. Professional bodies in the UK and internationally have recommended that genomic tests are not provided directly to consumers for the testing of asymptomatic children for adult-onset conditions for which no intervention can be made during childhood. *The Government should consider which, if any, genomic tests for asymptomatic children should be able to be provided directly to consumers, including whether there should be a ban on the provision of genomic tests for use on children that do not meet the criteria of the UK National Screening Committee.* (Paragraph 95)

15. Prenatal genomic testing could influence decisions on terminating fetuses, leading to specific concerns in addition to the issues concerning other types of direct-to-consumer genomic testing. *The Government should consider if any restrictions should be placed on the conditions that prenatal genomic tests provided directly to consumers are able to test for.* (Paragraph 99)

16. *The Government should consider requiring any manufacturer making genomic tests available to consumers in the UK to register a legal representative in the UK, with responsibility for ensuring that products supplied to consumers in the UK meet all relevant UK regulatory requirements.* (Paragraph 101)

17. As well as companies offering products to consumers that combine genomic testing with analysis of the genomic data obtained through the test, some companies offer secondary analysis of genomic data obtained through a previous genomic test from a different company. There is a potentially increased risk that the testing process used to obtain the genomic data is unsuitable for the analysis performed by a company offering secondary analysis. Despite this, the current regulation may not apply to companies providing secondary analysis of genomic data if their product is not deemed to include the use of physical equipment. *The Government should consider extending the definition of products covered by the regulation of genomic tests to include software and other services offering analysis and interpretation of genomic test results obtained from third parties.* (Paragraph 104)

Formal minutes

Thursday 3 June 2021

Members present:

Greg Clark in the Chair

Aaron Bell	Mark Logan
Dawn Butler	Rebecca Long-Bailey
Chris Clarkson	Carol Monaghan
Katherine Fletcher	Graham Stringer
Andrew Griffith	Zarah Sultana

First Report of Session 2021–22: *Direct-to-consumer genomic testing.*

After consulting all Members of the Committee, the Chair was satisfied that the Report represented a decision of the majority of the Committee and reported it to the House. (Order of the House of 24 March 2020).

Witnesses

The following witnesses gave evidence. Transcripts can be viewed on the inquiry publications page of the Committee's website.

Wednesday 17 June 2020

Dr Tara Clancy, Council Member, Nuffield Council on Bioethics — Q1–24

Graeme Tunbridge, Director of Devices, Medicines and Healthcare products Regulatory Agency — Q25–49

Lord Bethell of Romford, Minister for Innovation, Department of Health and Social Care; **Professor Sir Mark Caufield**, Chief Scientist, Genomics England; **Graeme Tunbridge**, Director of Devices, Medicines and Healthcare products Regulatory Agency — Q50–112

The following witnesses gave evidence during the Commercial genomics inquiry in the last Parliament. Transcripts can be viewed on the inquiry publications page of the Committee's website.

Tuesday 15 October 2019

Professor Anna Middleton, Chair, Association of Genetic Nurses and Counsellors; **Dr Matthew Hurles**, Head of Human Genetics and Senior Group Leader, Wellcome Sanger Institute; and **Professor Anneke Lucassen**, Chair, British Society for Genetic Medicine — Q1–54

Monday 28 October 2019

Kathy Hibbs, Chief Legal and Regulatory Officer, 23andMe; **Avi Lasarow**, Chief Executive Officer, DNAfit; and **Carla Newell**, Chief Legal Officer and Chief Risk Officer, Ancestry.com — Q55–183

Published written evidence

The following written evidence was received and can be viewed on the inquiry publications page of the Committee's website.

COG numbers are generated by the evidence processing system and so may not be complete.

1 23andMe (COG0002)

2 Bioindustry Association (COG0005)

3 Department of Health and Social Care (COG0006); (COG0009)

4 Genomics plc (COG0003)

5 Mavrommatis, Dr Yiannis (Programme Director MSc Nutrition and Genetics, St Mary's University) (COG0007)

6 Roche Products Ltd (COG0004)

7 UKCloud Ltd (COG0008)

The following written evidence was received during the Commercial genomics inquiry in the last Parliament and can be viewed on the inquiry publications page of the Committee's website.

1 23andMe (CGN0050); (CGN0079)

2 Academy of Medical Sciences (CGN0021)

3 Ancestry.com (International) (CGN0056)

4 Antenatal Results and Choices (CGN0075)

5 Association of Genetic Nurses and Counsellors (CGN0008)

6 Association of Medical Research Charities (CGN0028)

7 Atlas Biomed Group (CGN0029)

8 Biochemical Society (CGN0071)

9 BioIndustry Association (CGN0068)

10 Birmingham Women's and Children's NHS Foundation Trust (CGN0078)

11 British Pharmacological Society (CGN0027)

12 British Society for Genetic Medicine (CGN0030)

13 British Society for Histocompatibility & Immunogenetics (CGN0024)

14 Cancer Genetics Group (CGN0007)

15 CARE (Christian Action Research and Education) (CGN0054)

16 Christian Medical Fellowship (CGN0083)

17 Clinical Ethics and Law Southampton (CELS), University of Southampton (CGN0041)

18 Clinical Leads of NHS Regional Genetics Services (CGN0013)

19 Congenica Limited (CGN0046)

20 Curtis Rogers, Partner, GEDmatch.com (CGN0001)

21 Dante Labs SRL (CGN0018)

22 Department of Health and Social Care and the Department for Business, Energy and Industrial Strategy (CGN0053)
23 Don't Screen Us Out (CGN0034)
24 Down's Syndrome Association (CGN0085)
25 Dr Andelka Phillips (CGN0025)
26 Dr Elizabeth Ormondroyd (CGN0061)
27 Dr Elizabeth Tunbridge (CGN0031)
28 Dr Felicity Boardman (CGN0012)
29 Dr Lucy Frith (CGN0052)
30 Dr Pauline McCormack et al. (CGN0057)
31 Dr Peter Fotheringham (CGN0082)
32 Dr Peter MacCallum (CGN0033)
33 Dr Ron Zimmern (CGN0020)
34 Dr Susie Cooke (CGN0088)
35 EMBL-European Bioinformatics Institute (CGN0038)
36 EthicsAndGenetics (CGN0042); (CGN0087)
37 Everything Genetic Ltd (CGN0005)
38 Genetic Alliance UK (CGN0062)
39 Genomics England (CGN0070)
40 Genomics plc (CGN0048)
41 Illumina (CGN0063)
42 International Society of Genetic Genealogy (ISOGG) (CGN0010)
43 Macmillan Cancer Support (CGN0051)
44 Micropathology Ltd, University of Warwick (CGN0002)
45 Miss Martha Gutteridge (CGN0017)
46 Mr Darius Meadon (CGN0064)
47 Mr John Rainsbury (CGN0009)
48 Mr Robert Watkins (CGN0016)
49 MRC Human Genetics Unit, University of Edinburgh (CGN0006)
50 Mrs Colette Lloyd (CGN0032)
51 Mrs Debbie Kennett (CGN0073)
52 Ms Kavita Frary (CGN0065)
53 National Institute for Health and Care Excellence (CGN0067)
54 Nkaarco Diagnostics Ltd (CGN0058)
55 Nuffield Council on Bioethics (CGN0049)
56 Orig3n, Inc. (CGN0077)
57 P4ML Ltd (CGN0055)
58 PHG Foundation (CGN0023)

59 Prenetics International and DNAfit (CGN0035)

60 Professor Melinda Mills (CGN0044)

61 Professor Timothy Frayling (CGN0080)

62 Regional Genetics Laboratory (CGN0059)

63 Royal College of Physicians of Edinburgh (CGN0036)

64 Shelford Group (CGN0037)

65 Simon Davies (CGN0003)

66 Susan Hedley (CGN0043)

67 The Association of the British Pharmaceutical Industry (CGN0066)

68 The 'Mind the Risk' consortium (CGN0045)

69 The Royal College of Physicians and the Royal College of Pathologists (CGN0022)

70 The Royal Society (CGN0019)

71 UK Clinical Genetics Society (CGN0060)

72 UK Research and Innovation (CGN0069)

73 University of Oxford (CGN0026)

74 University of Strathclyde. Centre for Lifelong Learning. Genealogical Studies Postgraduate Programme (CGN0047)

75 Universty of Exeter (CGN0081)

76 Wellcome Genome Campus Connecting Science (CGN0040)

77 Wellcome Sanger Institute (CGN0039)

List of Reports from the Committee during the current Parliament

All publications from the Committee are available on the publications page of the Committee's website.

Session 2019–21

Number	Title	Reference
1st	The UK response to covid-19: use of scientific advice	HC 136
2nd	5G market diversification and wider lessons for critical and emerging technologies	HC 450
3rd	A new UK research funding agency	HC 7781
1st Special Report	Balance and effectiveness of research and innovation spending: Government and UK Research and Innovation Responses to the Committee's Twenty-First Report of Session 2017–19	HC 236
2nd Special Report	Commercial and recreational drone use in the UK: Government Response the Committee's Twenty-Second Report of 2017–19	HC 270
3rd Special Report	Work of the Biometrics Commissioner and the Forensic Science Regulator: Government Response to the Committee's Nineteenth Report of Session 2017–19	HC 1319
4th Special Report	A new UK research funding agency: Government Response to the Committee's Third Report of Session 2019–21	HC 1363
5th Special Report	5G market diversification and wider lessons for critical and emerging technologies: Government Response to the Committee's Second Report of 2019–21	HC 1377