Written Evidence Submitted by 23andMe

(COG0002)

Introduction

- 23andMe is a personal genetics and biotechnology company based in Mountain View, California, USA, that launched its Personal Genome Service (PGS) in the UK in December 2014
- 23andMe is the only US Food and Drug Agency (FDA) authorised direct-to-consumer genetic test in the world, with 10 million customers genotyped around the world
- 23andMe's PGS is an easy to understand DNA testing service that provides information for individuals to learn about their DNA, and to understand and benefit from the human genome
- 23andMe welcomes the Science and Technology Committee inquiry into commercial genomics
- This submission combines previous evidence submitted in person and in writing (alongside our latest company data) which focuses on our expertise of returning results to users across the globe – selling 250,000 kits in the UK and with 10 million genotyped customers globally

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

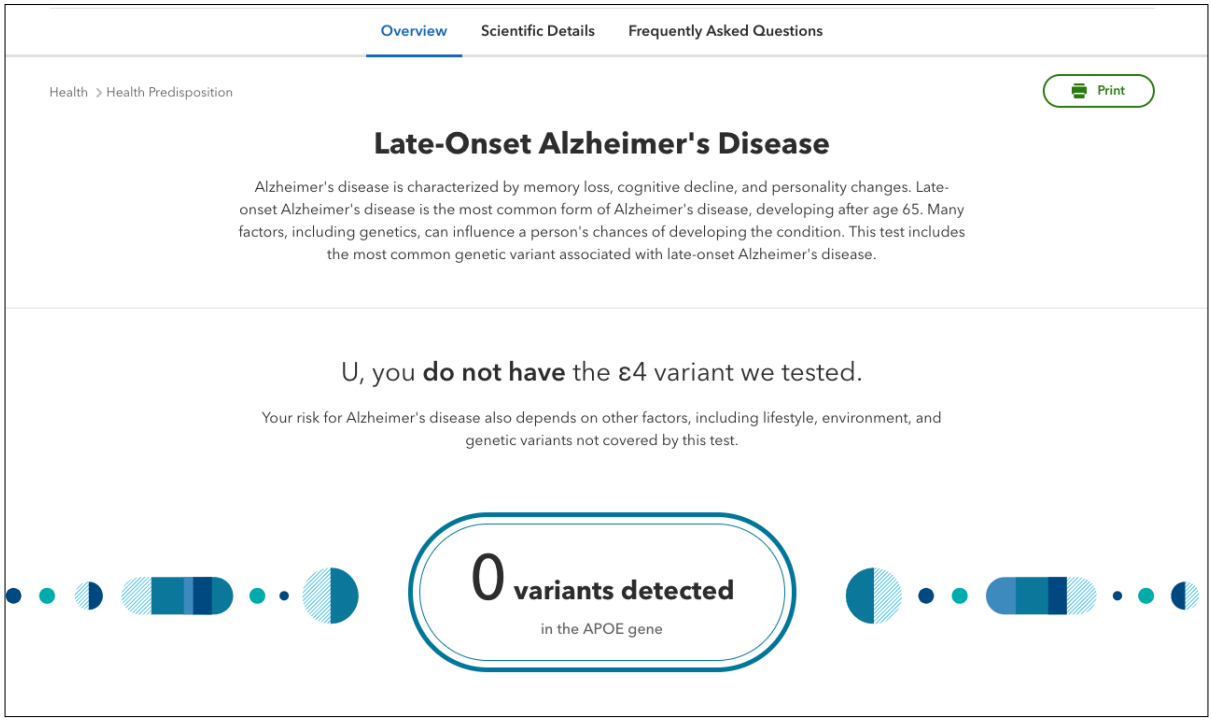
1.1. 23andMe's PGS gives people in the UK the opportunity to find out about their own genetic make-up.[1] This information enables individuals to be empowered to be involved in their own health and wellbeing and make conscious, more informed lifestyle choices

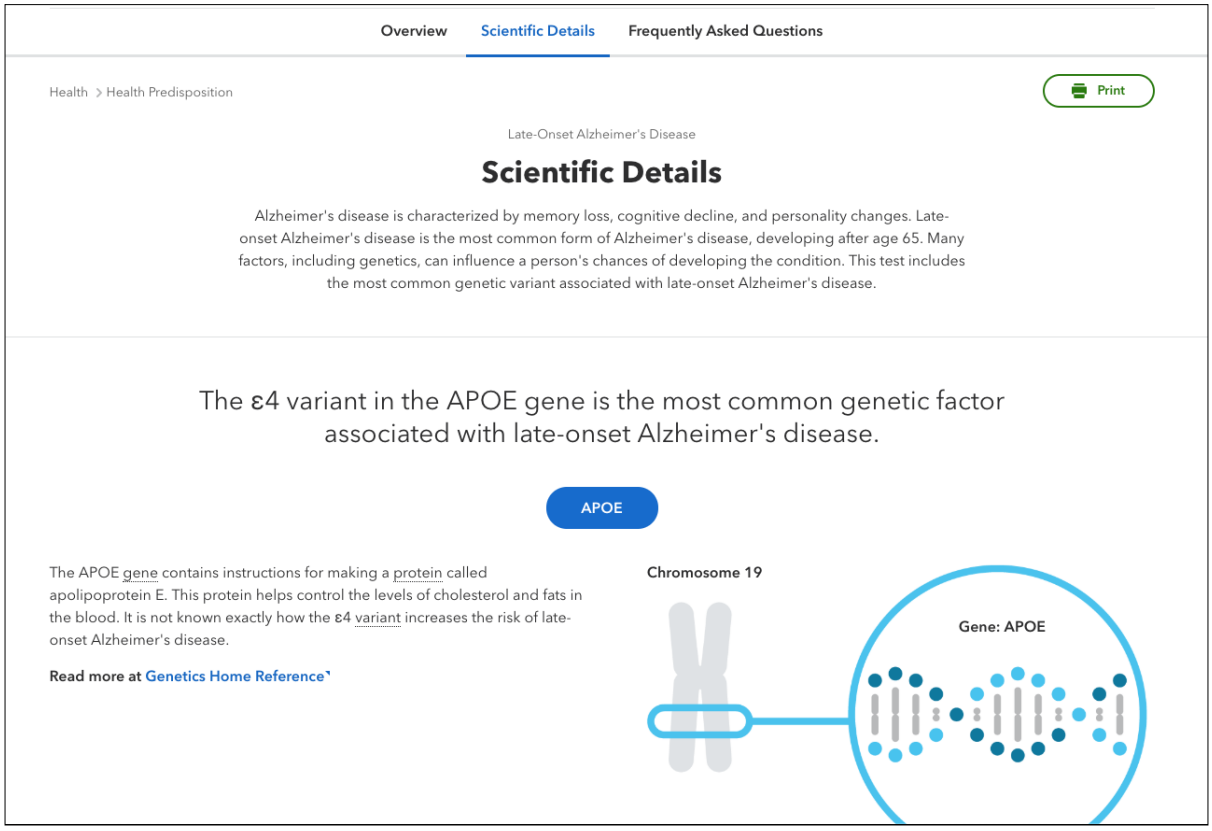
1.2. Through 23andMe, consumers are provided with a genetic test with labelling that has been clinically well-validated;[2] and is designed and validated for consumer comprehension. As a result, the 23andMe PGS test promotes greater genetic literacy and understanding of genetic health risk factors (see Figure 1)

1.3. *Figure 1: Example Genetic Health Risk Report*

[1] 23andMe, *Test Information,* available at: https://www.23andme.com/en-gb/test-info/

[2] 23andMe, *Genetic Science,* available at: https://www.23andme.com/en-gb/genetic-science/





2. The industrial strategy opportunity for genomics within the UK biotechnology sector, and how the Government could support UK growth (including for exports)

2.1. The genomics sector in the UK continues to grow, as a key component of the world leading life sciences sector, with the UK retaining its place as the global genomics leader. 23andMe has contributed greatly to the growth of this sector, with currently just over 250,000 UK customers. However, to capitalise on the world-leading position and to realise the potential of genomics, a supportive environment for the growth of the UK genomics industry must be maintained

2.2. The Life Sciences Industrial Strategy has provided a strong basis for the continued support of the sector. The second Sector Deal highlights the importance of genomics in delivering healthcare and the world leading position of the UK

2.3. In order to reach its ambitious targets of sequencing 5 million genomes, creating a network of genomic volunteers and increasing the use of genomic testing across the health service, it is important that the UK is a welcome environment for the genomics industry, fully utilising their expertise and innovations to achieve these goals

2.4. We urge the Committee to make enquiries to the Government and NHS England on their plans to continue supporting the genomics sector with the implementation of the NHS Long Term Plan

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

3.1. Whilst considering this submission, it is important that the Committee notes that 23andMe's technology is genotyping, rather than genomic sequencing and therefore what is detected by the PGS are well studied, peer reviewed health risk associations and not mutations that are suspected of being pathogenic

3.2. The 23andMe PGS is the only US FDA approved direct-to-consumer genetic test in the world. The reports are not intended to diagnose any disease, tell users anything about their current state of health, or to be used to make medical decisions, including whether or not users should take a medication or how much of a medication users should take

3.3. The 23andMe PGS has established analytical criteria for the detection of genetic variants in a manner that demonstrates consistent, accurate results for variants associated with carrier status and genetic health risk for certain disease areas

3.4. The test has a high level of analytical validity – it has demonstrated 99% concordance with Sanger sequencing, which is considered the clinical "gold standard," and 99% precision.[3] Test results are returned to users in an easy-to-understand format with users demonstrating 90% or higher comprehension of our results in multiple studies[4]

3.5. Our PGS provides 125+ online genetic reports[1] to customers, including:

- 9 genetic health risk status reports
- 40+ carrier status report
- 8 wellness reports
- 30+ traits

[3] ScienceDirect, *Sequencing,* available at: https://www.sciencedirect.com/topics/biochemistry-genetics-and-molecular-biology/sequencing

[4] 23andMe Personal Genomics Service, *Genetic Health Risk package insert,* available at: https://permalinks.23andme.com/pdf/PN-20-0279.pdf

- 35+ ancestry reports

3.6. When users purchase a 23andMe testing kit, they are provided with a package insert which provides details on intended use, important warnings and limitations, test performance, user studies and specific test information[5]

3.7. For a genetic association to be included in a curated Health report, there must be multiple sources of scientific evidence supporting the link between the genetic variant and the underlying phenotype. 23andMe also look for functional and biological evidence supporting a causative role for the variant's effect on the condition or trait. There must also be no compelling contradictory evidence refuting the genetic association. Taken together, the evidence should establish a consensus that the variant has a meaningful and real effect on the condition of interest

3.8. All 23andMe Health test results are returned to users in an easy-to-understand format which has been shown to be comprehensible by average consumers at a rate of at least 90% per report concept in multiple studies.[6] In addition each Health report category includes an educational module which is intended to further support the safe and effective use of the 23andMe test results

3.9. Six user comprehension studies were performed to assess how well people understand the PGS Carrier Status test reports. Studies are performed by an independent firm with expertise in recruiting for and performing general population studies using quota-based sampling to ensure that participants represented a wide range of demographic characteristics and were not current or prior customers of 23andMe. 23andMe has conducted six separate user comprehension studies, covering all key concepts in its health reports. In the six studies there were 2,239 demographically diverse participants: by gender (1,251 female and 988 male); by race (1,332 Caucasian, 329 African, 167 Asian, 351 Hispanic and 60 mixed race), by age (716 between the ages of 18-34, 795 between the ages of 35-54 and 728 who were 55 or older) and by level of education (732 having completed secondary education or less, 640 having attended some college and 867 having completed college and/or post graduate studies). User comprehension does not vary in any significant way between participants in any of the above demographics

4. The counselling or other support offered for those receiving, or considering asking for, commercial genomic test results, and whether this is to the standard required

4.1. The 23andMe PGS test includes health predisposition and carrier status reports

4.2. The test uses qualitative genotyping to detect select clinically relevant variants in the genomic DNA of adults from saliva for the purpose of reporting and interpreting genetic health risks and reporting carrier status. It is not intended to diagnose any disease

4.3. There are many things for a user to think about when deciding whether genetic testing is right for them. Although these tests can provide important information about health risks, they can also be upsetting or raise questions about what the results mean

4.4. Pre-purchase information[7] is available on our public website to assist a potential user in making an informed purchase. This information is as follows:

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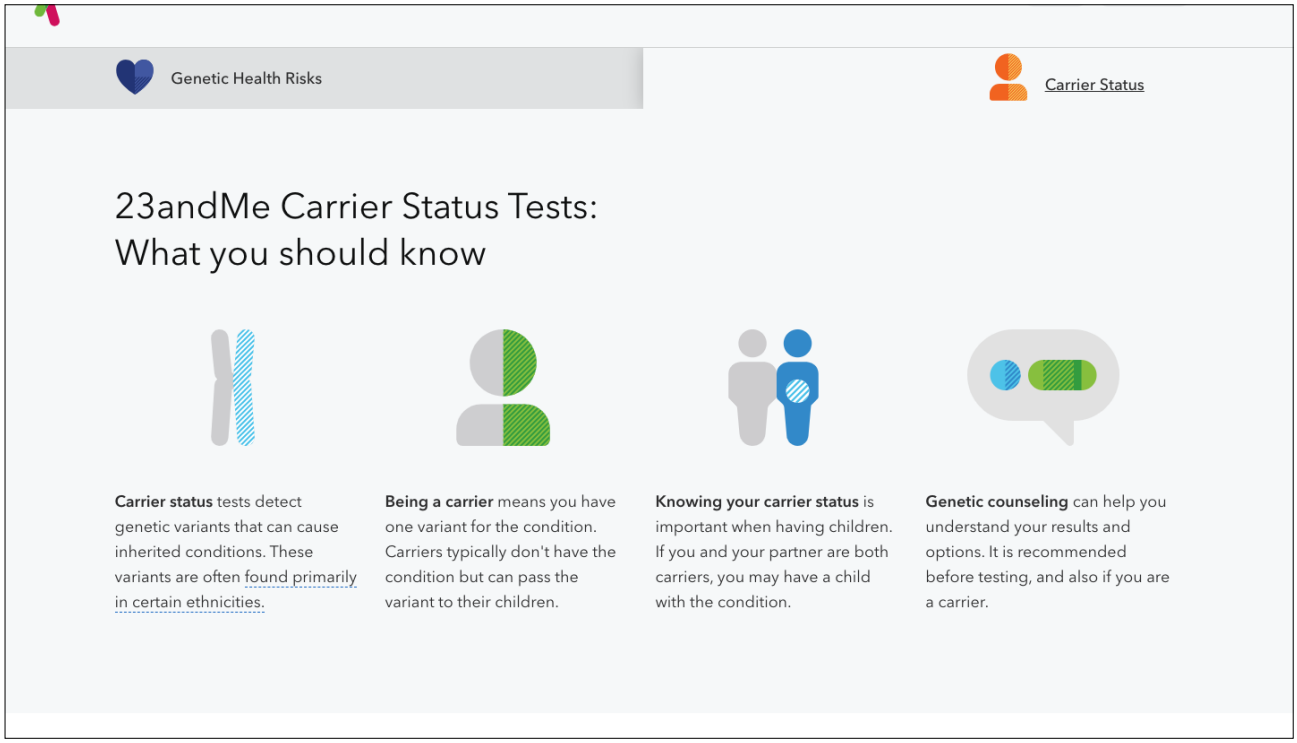
[5] 23andMe, *Genetic Health Risk Reports V5 Package Insert,* available at: https://permalinks.23andme.com/pdf/PN-20-0279.pdf

[6] 23andMe Personal Genomics Service, *Genetic Health Risk package insert,* available at: https://permalinks.23andme.com/pdf/PN-20-0279.pdf

4.4.1. Carrier Status Tests (CSTs)[8]:

- CSTs detect genetic variants that can cause inherited conditions. These variants are often found primarily in certain ethnicities
- Being a carrier means having one variant for the condition. Carriers typically don't have the condition but can pass the variant to their children
- Knowing carrier status is important when deciding to have children. If a user or their partner is a carrier, they may have a child with the condition
- Genetic counselling can help users understand their results and options. It is recommended before testing, and also if a user is a carrier

4.5. *Figure 2: 23andMe Carrier Status Tests – what you should know*



4.5.1. Genetic Health Risk reports (GHRs)[9]

- GHR reports tell users about genetic variants associated with increased risk for certain conditions. They do not diagnose cancer or any other health conditions or determine medical action
- Having a risk variant does not mean users will definitely develop a health condition. Similarly, users could still develop the condition even if they don't have a variant detected. It is possible to have other genetic risk variants not included in these reports

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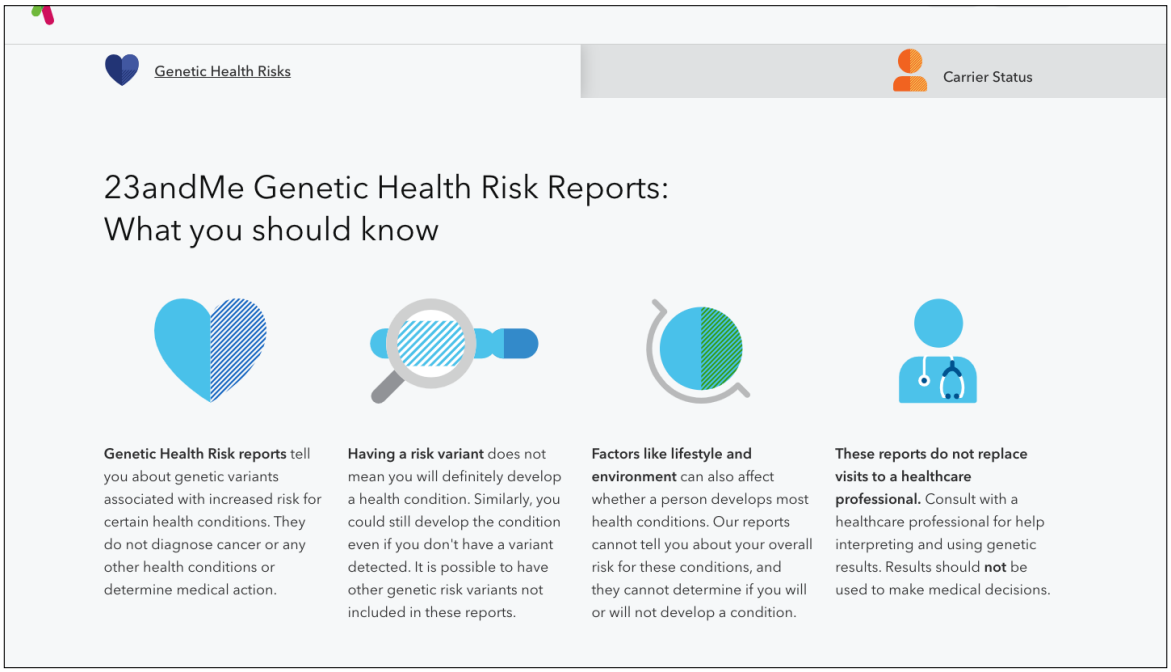
[7] 23andMe, *Health and Ancestry report,* available at: https://www.23andme.com/en-gb/dna-health-ancestry/
[8] 23andMe, *Sample Report: Carrier Status,* available at: https://permalinks.23andme.com/pdf/samplereport_carrierstatus.pdf
[9] 23andMe, *Sample Report: Genetic Health Risk,* available at: https://permalinks.23andme.com/pdf/samplereport_geneticheath.pdf

- Factors like lifestyle and environment can also affect whether a person develops most health conditions. Our reports cannot tell users about their overall risk for these conditions, and they cannot determine if a user will or will not develop a condition
- These reports do not replace visits to a healthcare professional. Consult with a healthcare professional for help interpreting and using genetic results. Results should not be used to make medical decisions

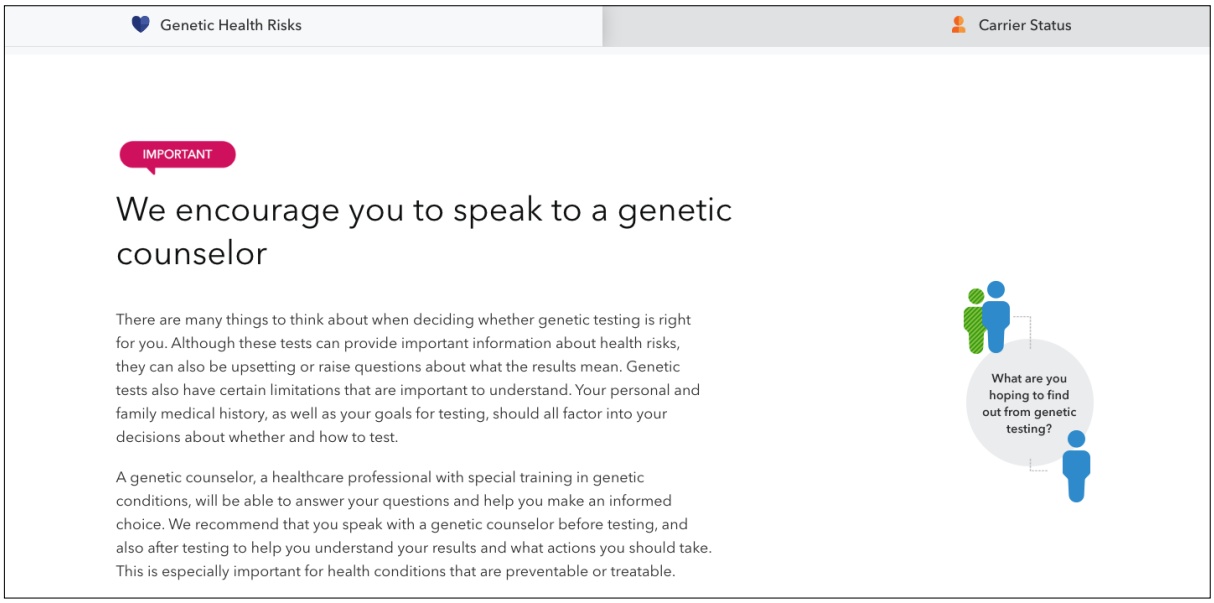
4.6. *Figure 3: 23andMe Genetic Health Risk Reports*



4.6.1. Genetic counselling

- There are many things for a user to think about when deciding whether genetic testing is right for them. Although these tests can provide important information about health risks, they can also be upsetting or raise questions about what the results mean. Genetic tests also have certain limitations that are important to understand. A user's personal and family medical history, as well as their goals for testing, should all factor into their decisions about whether and how to test
- A genetic counsellor, a healthcare professional with special training in genetic conditions, will be able to answer users' questions and help them make an informed choice. We recommend 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. This is especially important for health conditions that are not preventable or treatable

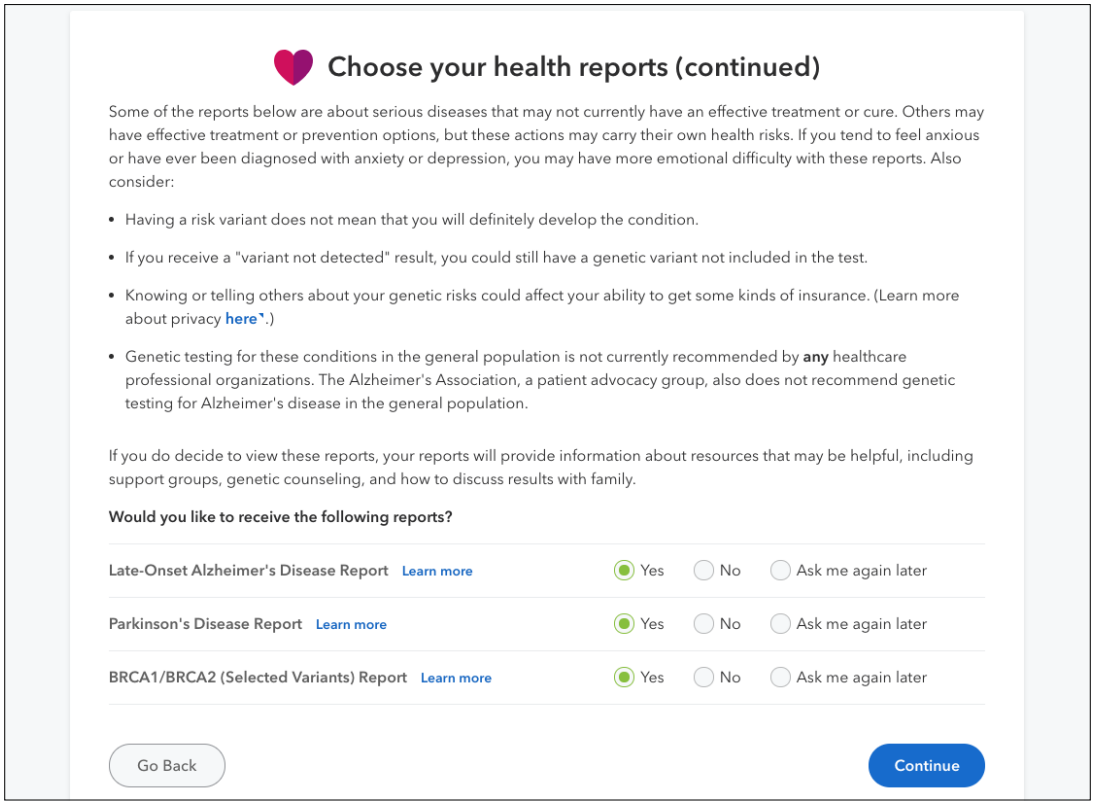
4.7. *Figure 4: 23andMe genetic counsellor information*



4.8. 23andMe's reports have been developed and tested to ensure that information is returned to users in an easy-to-understand format – comprehension studies show 90% overall comprehension of key result concepts

4.9. For certain reports (see Figure 5), customers are asked if they want to receive results and can choose to exclude some reports individually before results are returned

4.10. *Figure 5: Information provided to help customers choose their health reports*



4.11. Some of our reports are about diseases (including Alzheimer's) that may not have an effective treatment or cure. For these, customers must specifically opt-in to view the reports (default is opt-out). Customers can also choose to opt-in at a later date

4.12. 23andMe continues to work closely with UK agencies (for example, the Human Tissue and Embryology Authority) to ensure UK-focused signposting and resources are available to customers, and this will be launched shortly on the UK site

5. The potential benefits and risks for the NHS that arise from the increasing availability of commercial genomic testing

5.1. 23andMe's commercial genomic testing allows people to understand their own genetic make-up, including genetic health risks and wellness reports. This empowers individuals to be involved in their own health and wellbeing and make conscious, more informed lifestyle choices

5.2. Genomic data provides a step-change in the research potential of scientific and academic communities, and expanded commercial testing allows for larger cohorts of data. Around 85% of 23andMe UK users consent to their data being used for research, with participants answering online survey questions and researchers linking these to their genetic data to study traits and disease. These contributions help drive scientific studies. 23andMe participant data is de-identified and users can choose not to consent or to opt-out at any time

5.3. A common concern with expanding commercial genomic testing (including when 23andMe first launched in the UK) is an increased demand on the UK's health service. However, analysis of UK users undertaken in 2019 found that 1.9% had a specific conversation with their general practitioner (GP) about their report (in 2015, 4% of UK users had such a conversation with their GP). This consistency in action would reflect the likelihood of receiving actionable health reports. The 2019 analysis also highlighted that the format in which 23andMe returns results to users enables UK customers to understand the reports on their own and go to the doctor only when necessary

5.4. Prior to the launch of the 23andMe PGS in the UK, the MHRA discussed the 23andMe PGS with the US Food and Drug Administration (FDA) and shared information under the confidentiality commitment that is in place between the two agencies

5.5. Following the launch of 23andMe PGS in the UK, the company conducted a year-long voluntary post-market surveillance programme at the request of the 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 NHS

5.6. We have observed increased consumer involvement in their own healthcare, as a result of purchasing a 23andMe PGS, which can support public health objectives. It can also improve quality of life by enabling early identification of individuals at increased risk for various heritable conditions, thereby allowing targeted surveillance and management

5.7. Researchers have increasingly found engagement benefits resulting from consumer genetic testing and have also demonstrated that perceived risks have not materialised. In one such study, 93% of respondents indicated that getting a DTC test was "the right decision" for them, with 94% saying they would make the same decision again. Just 2% indicated that they regretted their decision to pursue testing[10]

[10] Public Health Genomics, *Direct-to-consumer Genetic testing: user motivations, decision making and perceived*

5.8. The impact of commercial genomic testing goes beyond directly improving patient outcomes. It is also essential to research; 23andMe sees individuals as research partners and so offers extensive opt-in opportunities for research. To date nearly 2 billion research questions have been answered by 23andMe's genotyped customers globally

5.9. It is important that healthcare professionals are appropriately trained, to reflect the advances in genomic technology, including DTC testing. 23andMe has repeatedly engaged with relevant UK stakeholders to provide updates on their technology

5.10. 23andMe welcomes the findings of Health Education England's Topol Review in relation to training healthcare professionals about genomics and increasing the genomic workforce. 23andMe would be delighted to further share our expertise in this area. We would urge the Committee to look into the recommendations of the Topol Review and determine how they will be implemented across the NHS

6. What data obtained from genomic testing could be used for if sufficient protection is in place for consumers using commercial genomic tests

6.1. 23andMe customers choose whether to participate in research. Participation requires an explicit and separate opt-in. Customers can withdraw their consent at any time, and choosing not to participate in research does not affect access to our service

6.2. When customers choose to participate in 23andMe research, they agree to a general research consent[11] for their de-identified data within the data aggregate (i.e., to be included among other participants' data) to be used in research analysis, internally and with collaborators. Customers can also choose to agree to an individual research consent[12] for their individual-level, de-identified data to be used in analysis, internally and with collaborators. Our collaborators include commercial companies, non-profit foundations and academic institutions

6.3. Research protocol and consent documentation are approved by an external institutional review board (IRB) to ensure our program meets the highest ethical and legal standards

6.4. Approximately 82% of our UK customers consent to research, and among UK customers who have joined 23andMe in 2020 the consent rate is approximately 86%. The number of customers who are participating in research and the amount of data each is providing have allowed 23andMe to gather together large cohorts (see Figure 6) to study some of the most common diseases

6.5. In April 2020, we launched a COVID-19 study of 23andMe's users. This is a longitudinal genome-wide association study with the aim of identifying genetic variants associated with differences in the severity of COVID-19. By identifying genetic variants that are more common in people who experienced severe disease, our scientists may be able to better identify who are the individuals most at risk. Perhaps even more importantly, these genetic studies can also help us gain new insights into how the novel coronavirus infects our cells and impacts our bodies. In turn, those insights might give us clues to potential targets for new drugs or vaccines

6.6. *Figure 6: Cohort studies of UK consented customers, as of 27 May 2020*

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utility of results, available at: https://www.karger.com/Article/FullText/455006

[11] 23andMe, *General Research Consent,* available at: https://www.23andme.com/about/consent/

[12] 23andMe, *Individual Data Consent,* available at: https://www.23andme.com/about/individual-data-consent

Cohort Study	Cohort numbers (UK)	Confirmed control numbers (UK)
Parkinson's	273	124,241
APOE e4 carriers	50,733	151,816
Cancer (non skin)	3,650	123,320
Depression	30,356	94,818
Asthma	22,432	104,200
Psoriasis	6,226	118,679
Cardiovascular disease	19,653	115.057
Colorectal cancer	336	125,289

6.7. 23andMe is guided by the principle that every individual deserves a private and secure environment to access and explore their genetics

6.8. Transparency is critical to providing clear and comprehensive resources that educate customers about our data practices and enable them to make informed decisions. Individuals are in control of what they want to learn, and what they want to share. It is our responsibility to safeguard and secure customer data

6.9. 23andMe's PGS is designed with the customer at the center. 23andMe provide individuals with greater access to their genetic information, and has built a strong privacy and data protection structure to ensure they have control over their information

6.10. 23andMe protects users' data in five key ways:

6.10.1. **Meaningful choice:** 23andMe gives users control over how their anonymised genetic information is used

6.10.2. **Privacy by design:** 23andMe takes great care to design their product with privacy in mind. The personal information collected when users register and use the site (such as name and email address) is kept separate from any genetic information provided to reduce the likelihood that users could be identified. If a user opts into research, their genetic information is stripped of personally identifying information and is assigned a randomised research identification number

6.10.3. **Third party sharing:** 23andMe will not sell, lease or rent individual-level information to any third party, even for research purposes, without explicit consent from the costumer. 23andMe does share aggregate information with third parties in order to perform 23andMe business development, initiate research, send marketing emails and improve services

6.10.4. **Security:** 23andMe believes genetic information, as well as the systems put in place to protect it, deserve the highest level of security. 23andMe employs software, hardware and physical security measures to protect the computers where customer data is stored. We use robust authentication methods to access our systems. Personal information and genetic data are stored in physically separate computing environments, in line with industry standards

6.10.5. **Research:** Customers choose whether or not they would like to participate in 23andMe research. Research participation is voluntary and requires an explicit and separate opt-in, and customers can opt-out at any time. Individual-level information will only be shared with a third party for 23andMe research if a customer provides explicit consent

6.11. Customers manage their own data. 23andMe will not make decisions for individuals about when and how to share their information, without explicit consent, unless otherwise required by law

6.12. The Terms of Service[13] and Privacy Statement,[14] available to the consumers pre-purchase, provide consumers with information about how 23andMe maintains customer privacy and data protection. Also, the Terms of Service clearly specify that 23andMe collects and processes the following types of information:

6.12.1. **Registration information:** Information provided when registering and/or purchasing our services (e.g., name, email/phone, DOB, payment info)

6.12.2. **Genetic information:** Information regarding a customer's genotype generated through the processing of a sample, or otherwise provided to 23andMe

6.12.3. **Self-reported information:** Information provided by a customer about themselves, generally through surveys, forms, or features (e.g. health related info, personal traits, ethnicity, family history, etc.)

6.12.4. **User content:** Information, other than Genetic Information and Self-Reported information, generated by users and transmitted to or through 23andMe (e.g., data, text, photographs, messages, or material posted on forums)

6.12.5. **Web-behaviour information:** Information on how users use the 23andMe website. Collected through cookies, web beacons etc. (e.g., IP address, Browser type, Operating system, clickstreams, and page views)

6.13. 23andMe will not use personal information in the following ways:

6.13.1. We will not sell, lease or rent personal information

6.13.2. We will not share any data (genetic or non-genetic) with an insurance company or employer

6.13.3. We will not provide information to law enforcement or regulatory authorities unless required by law

6.13.4. We do not share customer data with any public databases

6.14. A regular concern raised with 23andMe is the fear of 'genetic discrimination' from insurance companies who access reports produced by companies such as 23andMe. 23andMe will not provide any person's data (genetic or non-genetic) to an insurance company, and we support the UK's Code on Genetic Testing and Insurance. We would recommend that the Committee ask the Department of Health and Social Care what plans are in place to increase public awareness of this important agreement

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

7.1. 23andMe is compliant with both IVDD and GDPR regulations and is actively working with a notified body to meet IVDR requirements

[13] 23andMe, *Terms of Service,* available at: https://www.23andme.com/en-gb/about/tos/
[14] 23andMe, *Full Privacy Statement,* available at: https://www.23andme.com/en-gb/about/privacy/

7.2. As outlined above, ahead of and after launching our PGS in the UK, we undertook extensive engagement with the MHRA. We believe that all new entrants to the market should have discussions about undertaking a similar exercise to ensure that tests are suitable for UK consumers

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

8.1. 23andMe welcomes the continued focus on genomics; we believe there is benefit in more people knowing about their own genetic make-up. However, the question whether the NHS pays for this and who provides the service is a matter for the NHS and UK Government

9. The extent to which the 'in-vitro diagnostic medical device' regulation will address concerns about the validity of genomic test results and the provision of counselling alongside such tests

9.1. Products used in DTC genetic testing services providing health-related information are regulated as *in vitro* diagnostic medical devices and are subject to European Union legislation that sets out minimum standards of safety and quality. 23andMe's PGS meets all these requirements and the product is currently classified as a lowest risk category (Class A/1) device.[15] We believe that the current regulation reflects the level of 'risk' associated with products. The company is actively working with a notified body to meet IVDR requirements

9.2. IVDR, for 23andMe's test would be a class C test; it would require a notified body third-party review, prior to being on the market, or to maintain it on the market. DEKRA is our notified body third-party reviewer. We have engaged with them and will have our ISO 13485 audit completed enough in advance that we expect our technical file review to be complete, and that we will be fully compliant with the IVDR by the deadline provided

9.3. The UK should make sure to harmonise whatever additional regulatory requirements it may put in place with those that exist through the IVDR, or in the US in similar regulations. Due to the global nature of genomic testing it is vital that there is harmony on regulation

(May 2020)

[15] European Union, *Regulation (EU) 2017/746 on in vitro diagnostic medical devices,* available at: https://eur-lex.europa.eu/legal-content/EN/TXT/HTML/?uri=CELEX:32017R0746&from=EN