Written Evidence Submitted by Lord Bethell, Parliamentary Under Secretary of State for Innovation (Lords)
(COG0009)

Dear Chair

Commercial Genomics Inquiry

Thank you for your letter of 25 June. It was a pleasure to give evidence to your inquiry and I am happy to respond to your further questions. I hope it is helpful that this letter also includes responses from MHRA and Genomics England to whom you wrote separately.

Genomics plays a central role in the life sciences sector. Companies that provide 'commercial' genomics products and services, ranging from genomic sequencing and testing to sophisticated genomic data analysis, form an integral part of a thriving life sciences industry here in the UK. The genomics sector more broadly is set to grow with anticipated future investment from both government and industry to support the UK's future resilience to pandemics and other health emergencies. It is now more important than ever that we continue to support UK companies to contribute to a strong, resilient and innovative life sciences industry across the UK, and enable them to take full advantage of new opportunities and support the post-COVID-19 recovery.

I have responded to the questions raised in your letter of 25 June immediately below. Please find also attached in **Annex A** responses provided by MHRA and Genomics England.

Question 1: Can you confirm that it is the Government's intention to introduce new legislation superseding the Medical Devices (Amendment…) (EU exit) Regulations 2019, and that the UK will not implement provisions of the 'in vitro diagnostic medical device' regulation?

Response: The In Vitro Diagnostic Medical Devices Regulation 2017/746 (IVDR) will not take effect until we are outside of the transition period (May 2022) therefore, it will not automatically apply in UK law. We are currently taking steps to plan for after the end of the transition period. All decisions will be taken with a view to prioritising patient safety and ensuring patient access for IVDs and will be subject to the outcome of future negotiations. The IVDR is based heavily on international standards

and global harmonisation, which will be a key consideration for us in any future system.

An amendment to the 2019 ‘no deal’ Regulations will be laid in the Autumn, which will include the existing regulations (MDR 2002) and measures to amend deficiencies caused by the UK leaving the EU. Work is underway to develop the future UK regulatory regime for devices ready for 1 January 2021 and will include comprehensive legislation on medical devices and In Vitro medical devices matching the scope of the UK Medical Devices Regulations (MDR) (2002).
The Government remains committed to reaching an agreement with the EU on our future relationship. We will, of course, ensure that our future regulatory system takes account of agreements reached during the negotiations.

Question 2: Representatives of 23andMe, Ancestry.com and DNAfit indicated to the previous Science and Technology Committee that they would be willing to consider contributing to the training of genetic counsellors in the NHS. You told us that you were aware of the offer, but not of “any formal response”. Please could you update us on what the Government has done to take forward this offer of support?

Response: It may be helpful to clarify that I was aware that the companies had made this offer in their oral evidence to the Committee. The offer is very welcome, however, as far as I am aware they have not formally approached the Department or Health Education England. HEE is 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.

Question 3: Has the Government conducted any independent assessment of the current or potential impact and costs of direct-to-consumer testing on the NHS? If so, what were the results of that assessment, and if not, when will this work be completed?

Response: You referred to the earlier response of my predecessor, Baroness Blackwood, in late January when she said that ‘the Government will work with the NHS and others to explore how to build the evidence base’.

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. 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. As Sir Mark noted, the evidence submitted by 23andMe to your inquiry indicates that only a very small percentage (1.9%) of 23andMe’s 192,000 UK customers sought to have a

specific conversation with their GP about their test report. This would suggest that the impact on the NHS is currently relatively small.

I hope this reply is helpful. I look forward to receiving the Committee's report.

LORD BETHELL

Annex A

Responses to further questions in the Committee's letter to Professor Sir Mark Caulfield, Genomics England

Question 1: During the session, you told us that the Accelerating Detection of Disease challenge would "reach 5 million UK citizens on an absolutely equitable basis". Please could you provide more information on how participants will be selected, how you define equitable access, and how it will be guaranteed?

Response: The Accelerating Detection of Disease (ADD) challenge is a research programme which aims to recruit up to five million participants into a world-leading research cohort to shed new light on the detection and treatment of common diseases, including cancer, dementia and heart disease. Access to the ADD will be equitable in that participation is voluntary and free of charge, and the programme is being designed to ensure accessibility to all, according to a robust research protocol. This will help ensure that results from the study are valid and can be used to inform future treatment strategies for the entire UK population.

The protocol will be agreed by the ADD Board and the ADD Ethics & Feedback Advisory Group. Participants in ADD will reflect the UK population as closely as possible. This is one of the fundamental principles and aims of the programme. Diversity is a priority and recruitment strategies will be designed to ensure representation from harder to reach communities and groups. The process by which this will be achieved is currently being designed. The plan is to test a number of different recruitment strategies as part of the pilot phase of the programme to make sure that it works for a broad range of participants. The development process includes significant work to co-design and involve diverse communities from the start, working with them to build the project in a way that will ensure it is accessible, appropriate, culturally sensitive, and competent.

Question 2: You also said that the criteria for which results to return had not yet been agreed. What did Genomics England learn from providing secondary results to participants in the 100,000 Genomics Project?

Response: For clinical care, Genomics England (GEL), in partnership with NHS England, prioritised findings directly relevant to the rare disease diagnosis or cancer care to maximise the value for the participants' health problem. GEL were in advanced preparations to feed back additional findings to the NHS when COVID-19 struck so do not yet have evidence of the impact or patient perspectives on receipt of these findings. As soon as it is possible to support participants via the NHS we will restart the additional findings feedback process.

Question 3: Will the consent process for the Accelerating Detection of Disease challenge be comparable to the process for the 100,000 Genomes Project, and will there be a Participant Panel for the challenge as there was for the 100,000 Genomes Project?

Response: The consent process for ADD is still in development, ahead of the start of the pilot phase of the programme. Further details will be available this autumn. ADD will have a Participant Panel once the programme goes live. During the development phase, ADD already has in place an initial Co-Design Panel comprising members of the public from a range of communities and backgrounds. This group has been providing detailed feedback that has been fed into the design of the information for participants.

Question 4: With regards to results from direct-to-consumer genomic testing being shared with Genomics England's databases, you indicated that many direct-to-consumer genomic tests do not meet the standard required to enable this. What standards or other criteria would direct-to-consumer tests have to meet in order for data-sharing to be a possibility?

Response: My evidence was referring to use of the data from 23andMe in direct healthcare in the NHS. 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. It is correct that many of these direct to consumer tests may not be generated within a clinical grade pipeline that meets EU standards. I understand that 23andMe has a US accredited laboratory pipeline but many of the genetic markers they measure are more relevant to research into complex diseases.

Question 5: Has the Government undertaken an independent review of the impact of direct-to-consumer genomic testing on demand for GPs, clinical genetic and other NHS staff? If so, what were the results?

Response: Please see above.

Responses to further questions in the Committee's letter to Graeme Tunbridge, Director of Devices, MHRA

Question 1: Firstly, you described the "software and databases that are used" as one of the areas that could benefit from regulatory review, noting that "it is essentially up to the company to determine some of those things" currently.1 When you asked for "more rigour" in the provision of information on this, were you referring simply to companies being more transparent in the software and databases they use or would you support the introduction of certain minimum standards for software and databases?

Response: Whilst the IVDR will not automatically apply after the UK leaves the EU, it remains a useful reference point for regulatory strengthening as it is based on international best practice. The rigour referred to by Graeme Tunbridge reflects the enhanced requirements for clinical evidence that the IVDR would bring to support the claims being made by the manufacturer on an IVD. A key component of the clinical evidence is the scientific validity of the test, which for genomic testing is the extent to which a mutation in a particular gene (or genes) correlates to a meaningful association with a clinical condition or predisposition to that condition. The reference database and bioinformatics software used by the company performing the testing are critical to the scientific validity of the test.

Unlike under the current legislation, the IVDR would require a higher degree of scrutiny to be applied to these devices before being placed on the market. The software would be assessed as part of the clinical evaluation of the device and thus the manufacturer would be required to demonstrate all the verification and validation testing performed on the software to support that the device is in line with its intended purpose. Transparency is important especially when considering the type of database used to evaluate or compare the results obtained from a genetic test, as well as the accessibility to such databases. Furthermore, transparency is crucial when considering the scientific evidence to establish a correlation between a genetic test and the validity of its results.

Question 2: Secondly, you indicated that there may be aspects of the IVDR that could be better-tailored to the UK's situation, mentioning in particular the situation with counselling. Are there any other particular examples that you would highlight?

Response: The IVDR is based heavily on international standards and global harmonisation, which will be a key consideration for the Government in any future regulatory system. We will also need to carefully consider aspects where we may wish to go further in order to better protect UK patients and to adapt international standards to our unique domestic healthcare system. The Medicines and Medical Devices Bill will enable the Government to make regulations that improve the standards of scrutiny of IVDs placed on the UK market. This includes operating a more effective, robust and transparent regulatory system, ensuring that all decisions will be taken with a view to prioritise patient safety and patient access to IVDs.

Considering aspects of the IVDR relating to genetic counselling, we would want to examine international best practice in this area to understand when and how this is needed. There is unlikely to be a one size fits all approach, but we recognise the importance of ensuring that individuals are provided with relevant information on the nature, significance and the implications of the genetic test.

(10 July 2020)